Management of

HYPERTENSIVE DISEASES

Management of

HYPERTENSIVE DISEASES

JOSEPH C. EDWARDS, A.B., M.D., F.A.C.P., F.A.C.C.

Assistant Professor of Clinical Medicine, Cardiovascular Consultant to Division of Gerontology, and Consultant in the Hypertension and the Cardiac Clinics, Washington University School of Medicine and Barnes Hospital, St. Louis, Mo.; Cardiologist and Director of Hypertension Clinic, St. Luke's Hospital, St. Louis, Mo.; Active Staff Physician, Deaconess Hospital, and Member of Consultant Staff, Missouri Baptist Hospital and St. Joseph Hospital, St. Louis, Mo.; Consultant, Council on Drugs, American Medical Association; Medical Consultant, Fifth Army of the United States, Office of the Surgeon, Chicago, Ill.

With Foreword by PAUL DUDLEY WHITE

ILLUSTRATED

THE C. V. MOSBY COMPANY ST. LOUIS 1960

Printed in the United States of America
Library of Congress Catalog Card Number 60-11903
Distributed in Great Britain by Henry Kimpton, London

DEDICATED TO

Virginia Virginia Lee Joseph
and Jonathan Edwards

Foreword

Two quotations from Shakespeare's *King Henry VI* may be aptly applied to medical problems and to none more than to hypertension. In the first scene of the second act a messenger said:

> *And many strokes, though with a little axe,*
> *Hew down and fell the hardest-timbered oak."*

which may be said to illustrate the wear and tear from high blood pressure over the years and the need to control it or, better still, of course, to prevent it. In the eighth scene of the fourth act a still more significant truth is uttered:

> *A little fire is quickly trodden out,*
> *Which, being suffered, rivers cannot quench."*

Someday we must learn to recognize the candidates for serious hypertension before it strikes and to protect them from it, even in its earliest stages, before it has had any harmful effect. We are still at the threshold of such preventive medicine and have not yet done much of anything either to mark the candidates or to apply prophylactic measures.

Meanwhile, however, much is known about the effects of hypertension and much has been done, though largely empirically, to check it or in part to control it not only by the radical measures of surgery (thoracolumbar sympathectomy), diet (very low salt intake as in the rice diet), and the more powerful hypotensive drugs but also by the application of relatively simple hygienic measures of diet regulation, control of obesity, proper proportions of rest and exercise, and avoidance of excessive nervous strain and excessive use of tobacco and by the use of the milder sedative drugs.

This book by Dr. Edwards presents clearly and succinctly the present status of the problem of hypertension and its treatment. Important details of the use of the potent drugs alone and in combinations are included. As yet we are in the very midst of a rapid evolution of our knowledge not only of the treatment of hypertension but of that disease process itself. Without doubt new editions will be needed in the not far distant future to keep up with these advances.

Boston, Mass.

PAUL DUDLEY WHITE, M.D.

Introduction

The first American paper on hypertension was published by Janeway soon after the turn of the century and it stimulated an enormous number of clinicopathologic studies. Goldblatt more than anyone else opened the era of experimental work on hypertension about twenty five years later and when this century entered its third quarter drugs which could be depended upon to lower arterial pressure became available to the profession. To be sure they are not perfect drugs but at least they make a beginning. This book is a bold attempt to summarize the major developments in this exciting and fluid field. It should therefore be a useful guide to therapy.

Until the meaning of high blood pressure becomes apparent and more actuarial data are evaluated the manipulation of manometric readings will continue as the accepted procedure. It is however a maneuver which we must employ since there is certainly no evidence that a high arterial pressure is useful to the patient with progressive arterial disease. For the moment physicians must depend largely upon a variety of chemicals whose actions are complex sometimes unpleasant and always poorly understood. Empiric as these chemotherapeutic approaches may be there is reason to believe that they often relieve symptoms and sometimes prolong life.

Since there is no general agreement concerning the selection of drugs or the technique of their administration Dr. Edwards has faced a formidable job. He has wisely refrained from presenting criteria and schedules which are too rigid and yet the practitioner faced with a specific problem will find experienced counsel here.

THOMAS FINDLEY, M.D.

Professor of Medicine
Medical College of Georgia
Augusta, Ga.

Preface

This book is intended to serve as a guide to the practical management of the patient with hypertensive disease. It is written for the busy physician in practice who wants the latest information on the treatment of hypertension and guidance in selecting proper medication for a particular patient. A patient with mild arterial hypertension may be unable to respond normally to exertion or function tests depending upon the underlying disease. In more severe stages or forms of the disease the responses available are inadequate for even the ordinary demands of daily life.

A plan such as developed in this book should be followed to aid in a better understanding and a more logical use of the available agents in the management and control of hypertension. It is important to know what drugs to use and when to use them, but it is equally important to know what not to do.

Arterial hypertension may exist as idiopathic or essential hypertension caused by humoral or neurogenic factors or, perhaps more directly, as a condition secondary to acute or chronic renal disease. Hypertension secondary to pheochromocytomas, coarctation of the aorta, thromboses or strictures of the aorta or renal arteries is usually relieved when the cause is corrected surgically. Acute pyelonephritis and obstructive uropathy may be partially or temporarily reversible with specific therapy.

In the treatment of a particular patient the time may finally arrive when it is obvious to the physician that the therapy previously instituted is ineffective. The management of the disease then becomes more a matter of the skill and wisdom of the physician in the art of medicine rather than of medication alone. The patient himself and not his chemical environment becomes paramount. It is apparent that proper medical management before the onset of irreversible renal insufficiency will delay or even reverse the process underlying essential hypertension.

Frequently no explanation for the lack of correlation between height of blood pressure and symptoms or severity of the underlying lesion is available. A blood pressure which may be abnormal for a patient recently in good health may not necessarily be a defect requiring rigid control in the chronic "compensated" hypertensive patient. This is especially true in some patients in the older age groups who have a moderate elevation of blood pressure.

Thus interpretation both of the need for and the results of treatment often depends more upon the opinion of the clinician than upon the results of the laboratory tests alone. Each patient remains an individual to whom no one formula applies.

Although an understanding of the physiology and chemistry of the neurocirculatory system is important in the field of hypertensive disease, it is not within the scope of this book to go into detail about such mechanisms. However, a review of the experimental work and clinical studies of therapy in recent years has been included.

It is considered important to aid the clinician in keeping abreast of the recent developments in the therapy and management of the patient with hypertension by bringing together in one book many of the methods and observations a physician has found of value in his own experience.

The helpful criticism of Dr. Carl V. Moore, Dr. H. Mitchell Perry, and Dr. John R. Smith of the Washington University School of Medicine, who reviewed major portions of the manuscript for me, is gratefully acknowledged. Dr. Henry A. Schroeder gave many valuable suggestions. The kindness of Dr. Paul Dudley White and Dr. Thomas Findley, in whose laboratories much of my knowledge was acquired, in contributing the Foreword and the Introduction is deeply appreciated. The collaboration of Dr. Seymour Monat on the chapter on Hypertension Associated With Pregnancy is gratefully acknowledged. Acknowledgment is also made to the Washington University Department of Illustration for the photographs, to Dr. James N. Stokes for the discussion on the technique of aortography, and to the house staff of Barnes and St. Luke's Hospitals for capable assistance in the care of my patients. The clinic personnel were very cooperative. Dr. Paul O. Hagemann aided my interest in a hypertension clinic at St. Luke's Hospital.

The invaluable assistance of Miss Erma Frohbieter and Mrs. Minna Palfrey in typing and revising the material with tireless enthusiasm was of the utmost value. Miss Frohbieter helped also in reading the proof. Without the cooperation and forbearance of my wife, her help in typing the manuscript and in correcting the proof, and her deep understanding of the entire problem, this book could never have been finished. My entire family assisted me in spirit. Also I am grateful for the understanding attitude of my patients during the time this book was in preparation.

Such a book requires periodic revision; medical progress is a never-ending process. It is my hope that this material may aid in the better understanding and management of the patient with hypertensive disease.

JOSEPH C. EDWARDS, M.D.

Contents

chapter 1

Arterial Hypertension

chapter 2

Diagnosis of Arterial Hypertension

chapter 3

Heart Disease With Hypertension - - - - - - - - - - - 50

chapter 4

The Electrocardiogram in Hypertension - - - - - - - - 66

chapter 5

The Natural History of Essential Hypertension 86

chapter 6

Management of the Patient With Hypertension 103

chapter 7

Special Problems in Management of the Patient With Hypertension - - - - - - - - - - - - - - 153

chapter 8

Hypertension Associated With Pregnancy (Seymour Monat M D and Joseph C Edwards M D) - - - - - - 187

chapter 9

Special Types of Hypertension (Secondary Factors) 213

chapter 10

Antihypertensive Drugs—Pharmacology and Therapy 290

chapter 11

Etiology of Hypertension With Special Reference to Essential Hypertension 325

Management of

HYPERTENSIVE DISEASES



Arterial Hypertension

The medical and socioeconomic impact of hypertensive disease on American life is underscored by the knowledge that in 1955 49.5% of all natural deaths in the United States and 93% of the 810,200 deaths from cardiovascular disease were caused by arteriosclerosis and hypertension Of the three quarter million deaths in that year attributed to these two conditions 11% were caused by hypertension alone[1] About five million persons in this country have arterial hypertension of some degree if the upper normal limits for blood pressure are arbitrarily set at 140 mm Hg systolic and 90 mm Hg diastolic[2] It is difficult however to agree upon limits of normal blood pressure or to define levels that must be regarded as abnormal or as truly serious Within reasonable limits the critical factor in prognosis is not the mere height of the blood pressure measurement but more importantly the nature and degree of the underlying vascular disease Existence of elevated blood pressure may of course accelerate or promote other vascular pathologic changes present Until more is understood about the basic relationships and etiologies of vascular disease and arterial hypertension medical effort must be directed toward attempting safely to minimize diastolic pressures

DEFINITION

A definition of high blood pressure should not be based on any arbitrary level of pressure Instead frequency curves for an entire population should be studied together with selective group characteristics which might lead to a determination of susceptibles There should be follow up to identify the factors involved in appearance or progression of hypertensive disease in susceptible individuals[3]

Arterial hypertension represents a disorder which is characterized by an increase in systolic and diastolic pressure with normal cardiac output and by a general increase in peripheral vascular resistance This definition of necessity excludes elevated blood pressure resulting from coarctation of the aorta from inelasticity of the aorta and large arteries in the elderly individual and from arteriovenous shunts and increased blood pressure secondary to augmented cardiac output[4] The upper limits of blood pressure may be higher than the usually accepted values

Diagram vi–1

Head and upper extremities

Ductus Arteriosus

R Lung

S.V.C.

P.A.

Aorta

pul A.

L Lung

P.V.

pul V

Pulmonary Artery

L A

R.A.

L V

R.V.

I.V.C.

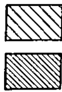
Descending Aorta

Trunk and lower extremities



Arterial blood (fully saturated)

Small admixture of venous blood
No visible cyanosis



Venous and arterial blood
Cyanosis visible

Venous blood

DIAGRAM VI-1

Tetralogy of Fallot with pulmonary stenosis before closure of the ductus arteriosus

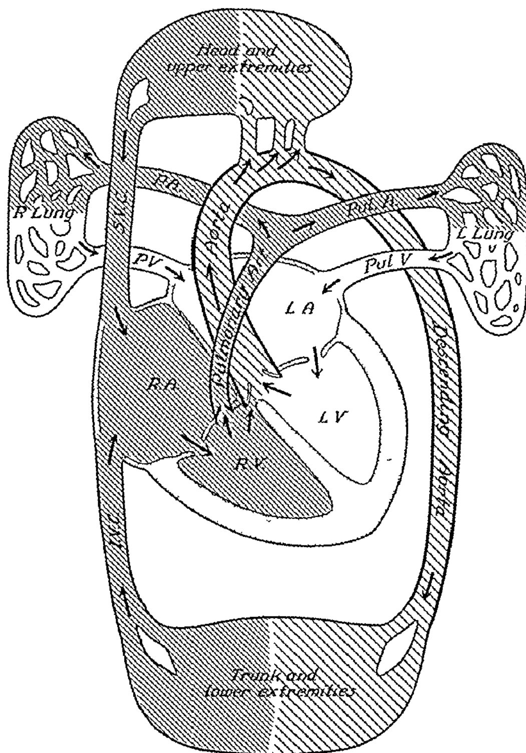
The four components of the tetralogy of Fallot are (1) pulmonary stenosis, (2) dextroposition of the aorta, (3) ventricular septal defect, and (4) right ventricular hypertrophy

The dextroposition of the aorta means that the aorta over rides the ventricular septum When this occurs the aortic septum cannot meet the ventricular septum in the normal manner hence the over riding of the aorta inevitably causes a high ventricular septal defect The foramen ovale closes normally There may, however, be a delay in the closure of the ductus arteriosus When this occurs the ductus arteriosus becomes of functional importance and increases the circulation to the lungs Under such circumstances the circulation of the blood is as follows

The blood from the right auricle flows into the right ventricle Part of the blood from the right ventricle is pumped out through the stenosed pulmonary artery to the lungs and part is pumped directly into the aorta All the blood in the pulmonary arteries goes to the lungs and the oxygenated blood is returned by the pulmonary veins to the left auricle thence it passes to the left ventricle The blood from the left ventricle is pumped out into the aorta Inasmuch as the aorta over rides the ventricular septum some of the blood from the right ventricle is also pumped directly into the aorta Therefore the aorta and the systemic circulation always receive a mixture of oxygenated blood from the lungs and venous blood from the right ventricle Most of the blood in the aorta is directed to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle Owing to the pulmonary stenosis and the low pressure in the pulmonary artery, some blood from the aorta flows through the ductus arteriosus into the pulmonary artery Thus the lungs receive blood from the right ventricle and also from the aorta through the ductus arteriosus The combined blood flow to the lungs means that a relatively large volume of blood reaches the lungs All the blood which reaches the lungs is returned by the pulmonary veins to the left auricle and thence to the left ventricle Consequently a relatively large volume of oxygenated blood is pumped out from the left ventricle This blood is mixed with the relatively small volume of blood which is pumped from the right ventricle into the aorta The resultant admixture of venous and arterial blood may be insufficient to produce visible cyanosis

Clinical diagnosis The heart is normal in size and in infancy there is only a systolic murmur Therefore the clinical picture at this age closely simulates that of a ventricular septal defect The electrocardiogram in addition to the usual right axis deviation, shows evidence of right ventricular hypertrophy

Diagram vi–2



Arterial blood (fully saturated)

Small admixture of venous blood
No visible cyanosis

Venous and arterial blood
Cyanosis visible

Venous blood

DIAGRAM VI-2

Tetralogy of Fallot with pulmonary stenosis after closure of the ductus arteriosus

The four components of the tetralogy of Fallot are (1) pulmonary stenosis, (2) dextroposition of the aorta (3) ventricular septal defect, and (4) right ventricular hypertrophy

The dextroposition of the aorta means that the aorta over rides the ventricular septum When this occurs it is impossible for the aortic septum to meet the ventricular septum in the normal fashion Thus dextroposition of the aorta inevitably means that there is a high ventricular septal defect uch is the nature of the ventricular defect in the tetralogy of Fallot In this malformation the foramen ovale closes and the ductus arteriosus undergoes normal obliteration

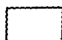
The blood which enters the right auricle flows into the right ventricle From the right ventricle part of the blood is pumped through the stenosed pulmonary orifice to the lungs where it is oxygenated and the oxygenated blood is returned by way of the pulmonary veins to the left auricle and thence passes to the left ventricle The blood from the left ventricle is pumped out through the aorta to the systemic circulation In asmuch as the aorta over rides the ventricular septum it thereby communicates directly with the right ventricle hence part of the blood from the right ventricle is directed out into the aorta Thereby the aorta and the systemic circulation receive blood from both the right ventricle and the left ventricle. All the blood in the systemic circulation is returned by the superior vena cava and the inferior vena cava to the right auricle and thence to the right ventricle Thus the right ventricle receives all the blood from the left ventri le and that part of the blood from the right ventricle which was pumped directly into the aorta. In contrast to this, the left ventricle receives only the blood from the right ventricle which was pumped through the pulmonary artery to the lungs Therefore the right ventricle always receives a greater volume of blood than does the left ventricle. Furthermore, the right ventricle has to pump the blood either through the stenosed pulmonary artery to the lungs or against systemic pressure. Hence the work of the right ventricle is increased and there results right ventricular hypertrophy

The right ventricle is enlarged and thick walled the left ventricle is small The two ventricles together give a heart of approximately normal size

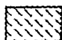
The pulmonary stenosis combined with the dextroposition of the aorta means that little blood reaches the lungs for oxygenation and that a large volume of venous blood is pumped into the systemic circulation Cyanosis is usually intense

Clinical diagnosis There is usually a history of attacks of paroxysmal dyspnea in infancy and of squatting in childhood The patient shows cyanosis and clubbing of the extremities The heart is of normal size A systolic murmur is the rule. The x ray shows an absence of fullness of the pulmonary conus The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

DIAGRAM VI–2


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

some blood flows from the right auricle to the left auricle The greater part of the blood in the right auricle flows into the right ventricle There the cycle starts again (see Diagram VI-3)

PHYSIOLOGY OF THE MALFORMATION

In the tetralogy of Fallot, regardless of whether there is pulmonary stenosis or pulmonary atresia, only a small volume of blood reaches the lungs for oxygenation, and a large volume of blood is pumped out through the aorta to the systemic circulation It follows that the pulmonary blood flow is reduced, the systemic blood flow is increased, and the volume of blood returned to the right auricle and the right ventricle is greater than normal The right ventricle not only has an increased volume of blood to pump, but it also encounters difficulty in the expulsion of blood through the stenosed pulmonary artery Furthermore, in many instances, in order to pump blood into the aorta the right ventricle must pump against systemic pressure It follows that the ejectile force of the right ventricle must be approximately the same as that of the left ventricle Nevertheless, since blood can escape from the right ventricle into the aorta, regardless of the severity of the pulmonary stenosis, the pressure in the right ventricle seldom greatly exceeds that in the left ventricle

The right ventricle undergoes slight dilatation and considerable hypertrophy As the foramen ovale tends to close, the strain on the right auricle is increased, it, too, may become hypertrophied The left ventricle, however, receives only a small volume of blood and therefore remains a relatively small chamber

The pulmonary stenosis breaks the ejectile force of the right ventricle and, in addition, it reduces the volume of blood which reaches the lungs Consequently the pressure in the pulmonary artery is normal or abnormally low and the expansion of the lungs is normal

Inasmuch as the patency of the ductus arteriosus is not primarily determined by mechanical factors, although the closure of the ductus arteriosus is often delayed for six months or even for one year, it is usual to find that, in spite of the pulmonary stenosis or even pulmonary atresia, the ductus arteriosus undergoes normal obliteration When this occurs in a patient with pulmonary atresia, life can be maintained only provided that there is sufficient collateral circulation to the lungs Under such circumstances the condition is functionally that of a pseudo truncus arteriosus in which the circulation to the lungs is by minute collateral vessels (see Chapter XIV) The clinical picture, however, closely resembles that of a tetralogy of Fallot with extreme pulmonary stenosis

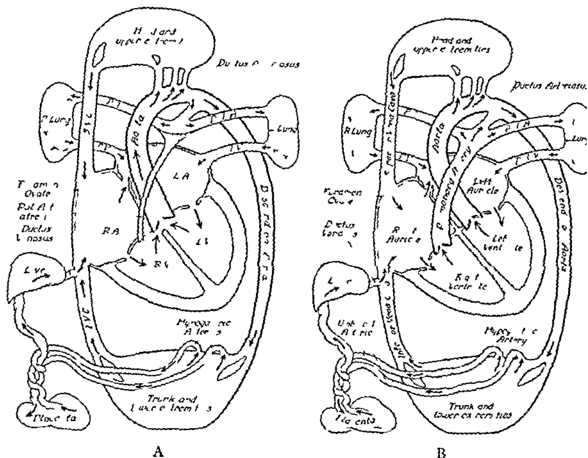


FIGURE VI-7 Fetal circulation (A) Tetralogy of Fallot with pulmonary atresia and (B) normal heart

frequently the only pathway by which the blood can reach the lungs Therefore even during fetal life blood must flow from the aorta through the ductus arteriosus into the lungs (see Figure VI-7) Since the lungs do not function and the pressure within the lungs is high, the pulmonary blood flow is very meager At birth the heart is frequently phenomenally small

Immediately after birth, with the expansion of the lungs, the pulmonary circulation is established by way of the ductus arteriosus The blood which flows from the aorta to the lungs is fully oxygenated in the lungs and is returned in the normal manner to the left auricle and thence to the left ventricle and the aorta All the blood in the aorta which is directed to the systemic circulation is returned by the superior vena cava and the inferior vena cava to the right auricle Consequently the volume of blood returned to the right auricle is greater than that returned to the left auricle and the pressure in the right auricle is higher than that in the left auricle The increased pressure in the right auricle tends to hold the foramen ovale open and therefore during the first days of life

Diagram VI-3

Tetralogy of Fallot with pulmonary atresia

When a tetralogy of Fallot is associated with pulmonary atresia and not pulmonary stenosis the course of the circulation is altered because no blood can flow through the atretic pulmonary artery to the lungs consequently the entire circulation to the lungs is by way of the ductus arteriosus Therefore unless extensive collateral circulation develops early the condition is compatible with life only as long as the ductus arteriosus remains patent.

Most of the blood from the right auricle flows into the right ventricle inasmuch as there is pulmonary atresia the blood cannot be pumped from the right ventricle into the pulmonary artery All the blood from the right ventricle is pumped out into the aorta, which over rides the ventricular septum and opens directly into the right ventricle. From the aorta the greater part of the blood flows into the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle Inasmuch as there is pulmonary atresia the pressure in the pulmonary artery is abnormally low and some blood from the aorta flows through the ductus arteriosus into the pulmonary arteries and thence to the lungs The oxygenated blood from the lungs is returned by way of the pulmonary veins to the left auricle Thence it passes to the left ventricle and is pumped out into the aorta where it mixes with the venous blood from the right ventricle. There the cycle starts again

The admixture of arterial and venous blood in the aorta combined with the relatively small volume of blood which reaches the lungs for oxygenation causes persistent cyanosis As the ductus arteriosus closes off less and less blood reaches the lungs, more and more blood is pumped through the systemic circulation Consequently the volume of blood which is returned to the right auricle becomes larger and the volume of blood to the left auricle becomes smaller As the pressure in the right auricle rises and the pressure in the left auricle falls, the valve covering the foramen ovale is forced open and the venous blood from the right auricle flows into the left auricle, thence it flows to the left ventricle and out into the aorta

Cyanosis becomes progressively more intense The obliteration of the ductus arteriosus usually renders the condition incompatible with life The duration of life and the amount of hypertrophy of the right ventricle depend upon the rate of obliteration of the ductus arteriosus

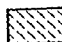
Clinical diagnosis Cyanosis is intense The onset of paroxysmal dyspnea occurs early in life The attacks may be long and severe and progress to loss of consciousness The infant gains weight slowly and may cease to gain The heart is small The murmur if present is systolic in time Diagnosis is based primarily on x ray findings Viewed in the anterior posterior position the heart is essentially normal in contour with no fullness of the pulmonary conus In the left anterior-oblique position the right ventricle is enlarged the left ventricle is relatively small and the pulmonary window is unduly clear, the heart is like a little round *apple* The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

DIAGRAM VI–3

Head and upper extremities
Ductus Arteriosus
R Lung
L Lung
S.V.C.
R.A.
PV
Aorta
Pul A.
Pul V
L.A.
R.A.
L.V.
R.V.
Strand of Pul Art
I.V.C.
Descending Aorta
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

Cyanosis usually becomes evident between three and six months of age. Often at first the infant suffers from attacks of cyanosis and paroxysmal dyspnea. Then after a number of such episodes cyanosis becomes persistent. Sometimes it is not until the infant begins to walk that cyanosis becomes apparent. In rare instances, even in childhood, cyanosis is apparent only upon exercise. Although the development of cyanosis is always distressing to the parents, the later its appearance, the less severe is the pulmonary stenosis. Therefore, when cyanosis is not suspected until early childhood, parents can be given the consolation that the prognosis is relatively better than if the cyanosis had developed at an earlier age.

The intensity of the cyanosis varies with the amount of available hemoglobin and the percentage of reduced hemoglobin in the circulating blood. The latter depends upon the degree of the pulmonary stenosis and the extent of the dextroposition of the aorta. After the closure of the ductus arteriosus, cyanosis is almost always present. If the stenosis is extreme, cyanosis appears early and becomes intense. Some patients show a purple glow over the entire body. Nevertheless, it is important to emphasize that many infants may suffer from severe degrees of anoxemia with only minimal cyanosis.

Polycythemia although seldom present in early infancy, is the rule in children and adults who suffer from anoxemia. It usually develops simultaneously with the development of the collateral circulation, and hence after the closure of the ductus arteriosus. The rate at which polycythemia develops depends upon the degree of oxygen unsaturation of the arterial blood and the demands of the body for oxygen. The red blood cell count, the amount of available hemoglobin, and the hematocrit reading usually remain within normal limits for the first six months of life. It is for this reason that cyanosis is so much less conspicuous in infants than in children. Indeed, only in patients with severe pulmonary stenosis is there marked polycythemia by two years of age.

In children and young adults the development of polycythemia is the rule. The red blood cell count is usually between 6 and 10 million and may occasionally reach 11 or even 12 million. The amount of available hemoglobin and the hematocrit reading are proportionally increased. The hemoglobin may reach 22 grams or 140 per cent. The hematocrit reading is usually between 60 and 80 per cent and in a few instances may rise above 90 per cent.

The blood platelets frequently become markedly reduced and the *blood fibrinogen* becomes abnormally low, as the blood becomes abnormally thick. Although the clotting time remains normal, the clot becomes extremely fragile.[10] These changes probably express an attempt on the body's part to compensate for

CLINICAL FINDINGS

The clinical findings vary with the severity of the pulmonary stenosis As previously mentioned, so long as the ductus arteriosus remains open the combined flow of blood to the lungs through the pulmonary artery and through the ductus arteriosus may be sufficient to prevent "visible" cyanosis

It is worthy of note that in Sandifort's case and in the first case reported by Peacock both physicians stressed the fact that the patient was perfectly normal at birth Furthermore, Sandifort postulated that the normality of color during early infancy was probably due to the flow of blood through the ductus Botalli!

Difficulty in feeding, failure to gain, and *stunting of growth* may be the first indications of an abnormality These difficulties result from an inadequate supply of oxygen The infant gains weight very slowly because he simply cannot digest much food Small and frequent feedings may help Many infants develop severe anemia, which adds to the difficulty *Cessation of weight gain* is a serious manifestation and is usually indicative of an extremely low arterial oxygen saturation During childhood the patient generally improves but growth is slow The onset of puberty may be delayed, nevertheless, the majority of individuals with a tetralogy of Fallot attain full physical development

Attacks of paroxysmal dyspnea are common in infants with a tetralogy of Fallot These attacks may be precipitated by nursing or by bowel movements, occasionally they occur without provocation The infant becomes very cyanotic, respirations are rapid, and there is often difficulty in expiration These attacks may progress to loss of consciousness and may even cause convulsions They apparently occur at the time when the ductus arteriosus is undergoing obliteration The period of greatest difficulty is usually between the ages of six and eighteen months From eighteen months to two years of age the infant holds his own During this time the attacks of paroxysmal dyspnea decrease and then cease, *thereafter the child starts to improve* For the treatment of paroxysmal dyspnea, see below and also Chapter V

Loss of consciousness is always a serious manifestation Infants seldom lose consciousness until the oxygen saturation of the arterial blood falls to 12 per cent It may fall even lower An extremely low arterial oxygen saturation should never be dismissed as a technical error, nor should it be assumed that the sample was taken from a vein, because the oxygen saturation of the arterial blood can be extremely low When the arterial saturation is only 10 or 12 per cent, there is real danger that a further fall may be fatal Indeed, such is the probable cause of sudden death, *which occasionally* occurs



FIGURE VI-8 Squatting

Although the habit of squatting is characteristic of children with a tetralogy Fallot, it is not limited to this malformation Children with a complete trans-sition of the great vessels combined with pulmonary stenosis also squat when ted The reason why these children squat is not known One child, who was n years of age, replied to the author s query as to why he squatted Can t you e? It cuts off the circulation to my legs and increases the circulation to my

the increased viscosity of the blood, they lessen the danger of thrombosis but increase the patient's tendency to hemorrhage

Telangiectases and *purpuric eruptions* frequently develop over the lower extremities in patients with long standing polycythemia Such eruptions are generally associated with dilatation of the capillaries and may be related to the changes in the clotting mechanism of the blood

Clubbing of the extremities develops as the red blood cell count rises It is the result of the compensatory polycythemia, the dilatation of the capillaries, and the persistent oxygen unsaturation of the arterial blood With extreme polycythemia the fingers and toes may show drumstick deformities After successful operation even pronounced clubbing may eventually disappear

Dyspnea on exertion is common The patient's ability to exercise may be extremely limited Although the heart rate increases with exercise, owing to the pulmonary stenosis the circulation to the lungs cannot be significantly increased The consequence is that, with an increase in the minute volume, the amount of blood which reaches the lungs remains constant and the increase in the heart rate causes more venous blood to be pumped into the systemic circulation There is a fall in the oxygen saturation of the arterial blood For a short period of time the increase in the heart rate may sufficiently compensate for the fall in the arterial oxygen saturation to maintain or even to increase the supply of oxygen to the body Nevertheless, the oxygen saturation of the arterial blood falls rapidly and to such a low level that the patient develops dyspnea and a corresponding increase in the intensity of the cyanosis

Squatting is a common habit among children with a tetralogy of Fallot (see Figure vi-8) These children are able to get their breath more easily in this position Sometimes the child runs for a short distance and then suddenly squats down The author had one patient who walked in a squatting position The benefit which may be derived by squatting was delightfully illustrated by another patient, who could play football but could not participate in any other sport Football was possible because, as he said, All the children ran and squatted down and ran again '

Children with severe pulmonary stenosis frequently desire to sit crouched on their legs They usually like to cross their knees and, if allowed, will double up the knees and sit leaning slightly forward with the chest upon the knees Most of these infants and children, when extremely fatigued, automatically assume the knee chest position, lying with the weight of the thorax on the shoulders and neck



FIGURE VI–8 Squatting

Although the habit of squatting is characteristic of children with a tetralogy of Fallot, it is not limited to this malformation Children with a complete trans position of the great vessels combined with pulmonary stenosis also squat when tired The reason why these children squat is not known One child, who was ten years of age, replied to the author s query as to why he squatted 'Can't you see? It cuts off the circulation to my legs and increases the circulation to my

the increased viscosity of the blood, they lessen the danger of thrombosis but increase the patient's tendency to hemorrhage

Telangiectases and *purpuric eruptions* frequently develop over the lower extremities in patients with long standing polycythemia Such eruptions are generally associated with dilatation of the capillaries and may be related to the changes in the clotting mechanism of the blood

Clubbing of the extremities develops as the red blood cell count rises It is the result of the compensatory polycythemia, the dilatation of the capillaries, and the persistent oxygen unsaturation of the arterial blood With extreme polycythemia the fingers and toes may show drumstick deformities After successful operation even pronounced clubbing may eventually disappear

Dyspnea on exertion is common The patient's ability to exercise may be extremely limited Although the heart rate increases with exercise, owing to the pulmonary stenosis the circulation to the lungs cannot be significantly increased The consequence is that, with an increase in the minute volume, the amount of blood which reaches the lungs remains constant and the increase in the heart rate causes more venous blood to be pumped into the systemic circulation There is a fall in the oxygen saturation of the arterial blood For a short period of time the increase in the heart rate may sufficiently compensate for the fall in the arterial oxygen saturation to maintain or even to increase the supply of oxygen to the body Nevertheless, the oxygen saturation of the arterial blood falls rapidly and to such a low level that the patient develops dyspnea and a corresponding increase in the intensity of the cyanosis

Squatting is a common habit among children with a tetralogy of Fallot (see Figure vi–8) These children are able to get their breath more easily in this position Sometimes the child runs for a short distance and then suddenly squats down The author had one patient who walked in a squatting position The benefit which may be derived by squatting was delightfully illustrated by another patient, who could play football but could not participate in any other sport Football was possible because, as he said, "All the children ran and squatted down and ran again"

Children with severe pulmonary stenosis frequently desire to sit crouched on their legs They usually like to cross their knees and, if allowed, will double up the knees and sit leaning slightly forward with the chest upon the knees Most of these infants and children, when extremely fatigued, automatically assume the knee chest position, lying with the weight of the thorax on the shoulders and neck

able to work eight hours a day and complained only of slight fatigue at nightfall even though he had an oxygen saturation of the arterial blood of only 54 per cent Indeed, he was so well adjusted that he desired operation only provided it would fit him for military service Inasmuch as this was not possible, operation was not undertaken

The blood pressure is often difficult to obtain because the pulse is weak and the pulse pressure is narrow The strength of the pulse, however, is equal in arm and leg

Systemic hypertension is relatively common in patients with a tetralogy of Fallot, especially when the pulmonary stenosis is extreme The etiology is not clear It may be secondary to the renal complications which frequently occur in patients with long standing polycythemia Elevation of the nonprotein nitrogen is relatively common Furthermore, it may return to a normal level after a successful anastomosis and the blood pressure may also become lower

Hemiatrophy is relatively common in patients with a tetralogy of Fallot When it does occur, the aorta almost invariably, if not invariably, arches toward the normal side of the body This phenomenon occurs so regularly that if a patient has a *left* hemiatrophy one can postulate that he has a *right* aortic arch

CARDIAC FINDINGS

The heart is usually strikingly small The malformation in itself places a constant load on the heart and does not cause progressive cardiac enlargement Therefore the heart usually remains within normal limits Indeed, in patients with a severe degree of pulmonary stenosis or atresia, the heart may be exceptionally small The right ventricular wall is, however, always increased in thickness and may become sufficiently enlarged to press against the anterior chest wall and cause left sided chest deformity

Murmurs are frequently absent during the first days of life and the infant is considered normal By the end of the first week, however, a murmur is usually audible It is *systolic* in time and of maximal intensity along the left sternal border It may be heard posteriorly but is usually not transmitted to the vessels of the neck Such murmurs are often better heard in the recumbent than in the erect position but occasionally may be heard only when the patient is leaning forward

The intensity of the murmur varies inversely with the severity of the pulmonary stenosis A mild pulmonary stenosis causes a loud systolic murmur, whereas if there is extremely severe pulmonary *stenosis* or pulmonary atresia, even though there is dextroposition of the aorta, no murmur is audible Conse

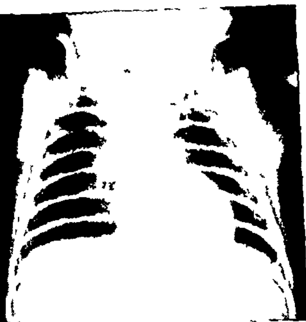
lungs" That may be the correct explanation Certain it is that these children get their breath more easily in a squatting position

The exercise tolerance of these children varies with the severity of the pulmonary stenosis and with the extent of the collateral circulation It also varies in the same individual from day to day Almost every child will say that 'on good days" he can do thus and so, and that 'on bad days" he can walk only a short distance or just a few steps Indeed, on very bad days he may be bedridden Extremes of heat and cold and damp weather lessen his exercise tolerance, dry air is a great boon Consequently the dry air at a high altitude may be better tolerated than the damp air at sea level

In some children with a tetralogy of Fallot exercise causes the oxygen saturation of the arterial blood to drop so rapidly that they can walk only a few steps before squatting to rest, whereupon the oxygen saturation of the arterial blood rises with equal rapidity and to such a height that there is no stimulus for the development of polycythemia Such patients have a normal red blood cell count and at rest may show no cyanosis Indeed, the color may be so normal that the family does not realize that the child is cyanotic The author knows of one instance in which the parents sought the aid of a psychiatrist because the child developed the habit of squatting Except for the absence of cyanosis, such children have a typical tetralogy of Fallot, hence the condition has been called an "acyanotic tetralogy of Fallot

The development of collateral circulation and that of polycythemia appear to go hand in hand As these increase, the oxygen saturation of the arterial blood rises The first indication of this may be a decrease in the number of attacks of paroxysmal dyspnea Later, as the child grows, his exercise tolerance usually increases He looks more cyanotic but he is stronger By the time the child reaches puberty he may be able to walk for considerable distances at a slow pace and generally no longer feels the need to squat This is due in part to the increase in the oxygen saturation of the arterial blood and in part to the fact that the collateral circulation develops from the systemic circulation Consequently an increase in the systemic blood flow tends to increase the flow of blood through the collateral vessels This results in an increase in the pulmonary blood flow

Over a period of years the oxygen saturation of the arterial blood gradually rises, so that most young adults have an oxygen saturation between 70 and 80 per cent at rest and show a less marked fall in the oxygen saturation upon exercise Even when the oxygen saturation of the arterial blood remains low it is remarkable how well these patients adjust The author had one patient who was



Anterior posterior position



Left anterior-oblique position

FIGURE VI-9 Tetralogy of Fallot with functional pulmonary atresia Infant

quently the intensity of the murmur offers a clue to the severity of the pulmonary stenosis

A systolic thrill is generally palpable if the murmur is moderately loud

The pulmonic second sound is usually faint but audible The small size of the pulmonary artery, combined with the small volume of blood which is forced through its orifice, renders the closure of the pulmonic valve difficult to hear Nevertheless, the aorta may be displaced so far to the left that the closure of the aortic valve is better heard to the left than to the right of the sternum Consequently, when the second sound to the left of the sternum is greater than the second sound to the right of the sternum, it is strong presumptive evidence of extreme dextroposition of the aorta

A third heart sound, or even a gallop rhythm, is seldom heard in patients with a tetralogy of Fallot The occurrence of a gallop rhythm is always indicative of a failing heart

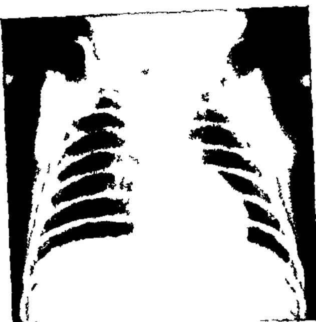
Cardiac failure is rare in patients with a tetralogy of Fallot Occasionally an infant with severe anemia and a tetralogy of Fallot may suffer from cardiac dilatation and cardiac failure Such infants develop engorgement of the liver but, owing to the inadequate blood flow to the lungs, these patients seldom suffer from pulmonary congestion

X RAY AND FLUOROSCOPIC FINDINGS

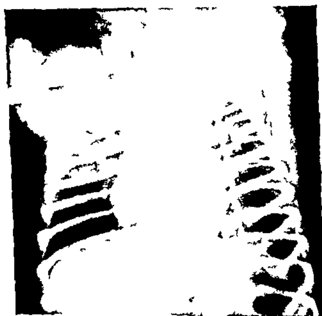
X ray and fluoroscopic findings vary with the age of the patient and the severity of the pulmonary stenosis

In infants with anatomical or functional pulmonary atresia the heart is frequently phenomenally small, as shown in Figure VI–9 (A–P position) and Figures VI–10 and 11 In the left anterior oblique position the enlargement of the right ventricle is usually apparent The heart has the appearance of a little round apple with a stem at the base This shape is caused by the enlargement of the right ventricle combined with a small left ventricle The absence of the pulmonary artery renders the pulmonary window abnormally clear and makes the aorta look like the stem of an apple, as shown in Figures VI–9, 12, and 13 (LAO position) When there is pulmonary stenosis, the heart is remarkably normal in size and shape (see Figure VI–14)

As the patient grows and the diaphragm descends, the heart assumes a characteristic contour In the anterior posterior position the heart appears to be normal in size but, owing to the stenosis of the infundibulum of the right ventricle, there is no fullness in the region of the pulmonary conus The upper margin



Anterior posterior position



Left anterior-oblique position

FIGURE VI-9 Tetralogy of Fallot with functional pulmonary atresia Infant



Figure VI–10 Tetralogy of Fallot with functional pulmonary atresia (same patient as in Figure VI–12) Infant

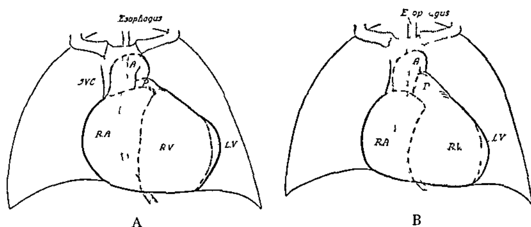


Figure VI–11 (A) Tetralogy of Fallot with functional pulmonary atresia and (B) normal heart Infant



Left anterior oblique position



Right anterior-oblique position

FIGURE VI-12 Tetralogy of Fallot with functional pulmonary atresia (same patient as in Figure VI-10) Infant



FIGURE VI-10 Tetralogy of Fallot with functional pulmonary atresia (same patient as in Figure VI-12) Infant

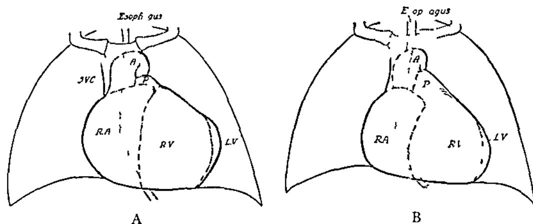


FIGURE VI-11 (A) Tetralogy of Fallot with functional pulmonary atresia and (B) normal heart Infant

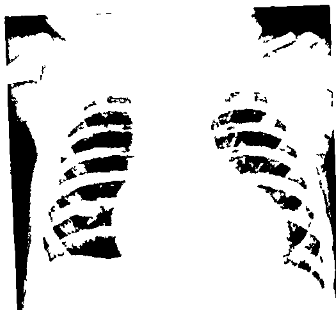
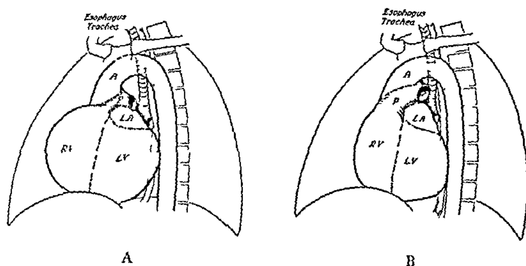


FIGURE VI-14 Tetralogy of Fallot with severe pulmonary stenosis (same patient as in Figure VI-1) Infant

of the cardiac silhouette to the left of the sternum, in contrast to the normal contour, is concave (see Figures VI-15 and 16)

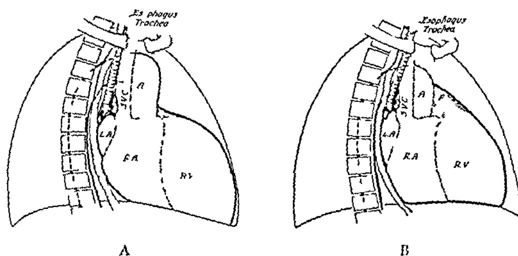
The small size of the pulmonary artery combined with the partial rotation of the great vessels causes the shadow cast by the great vessels to be narrow Consequently the contour of the heart is that of a *coeur en sabot* or 'boot shaped heart , this has been repeatedly emphasized by numerous observers[11 17] and is shown in Figures VI-17 and 18

A right aortic arch is relatively common in this malformation This condition is not to be confused with dextroposition of the aorta In the latter the aorta over rides the right ventricle, whereas the characteristic of a right aortic arch is that the aorta arches to the right Inasmuch as this variant occurs in approximately 25 per cent of all patients with a tetralogy of Fallot, the possibility should always be investigated (see Chapter XXVI) When the aorta arches to the right, the heart is usually long and narrow and its left border shows a slight concavity or a gentle slope (see Figures VI-19 and 20) The aortic knob is frequently visible to the right of the sternum within the shadow of the superior vena cava Even when the x ray or fluoroscopic examination does not show the aorta on the right, upon delineation of the esophagus by barium-opaque mixture, the esophagus is seen to lie to the left of the great vessels Under such circumstances,



A B

LEFT ANTERIOR OBLIQUE POSITION



A B

RIGHT ANTERIOR-OBLIQUE POSITION

FIGURE VI-13 (A) Tetralogy of Fallot with functional pulmonary atresia and (B) normal heart Infant



FIGURE VI-14 Tetralogy of Fallot with severe pulmonary stenosis (same patient as in Figure VI-1) Infant

of the cardiac silhouette to the left of the sternum, in contrast to the normal contour, is concave (see Figures VI-15 and 16)

The small size of the pulmonary artery combined with the partial rotation of the great vessels causes the shadow cast by the great vessels to be narrow Consequently the contour of the heart is that of a *coeur en sabot* or boot shaped heart", this has been repeatedly emphasized by numerous observers[11 17] and is shown in Figures VI-17 and 18

A right aortic arch is relatively common in this malformation This condition is not to be confused with dextroposition of the aorta In the latter the aorta over rides the right ventricle, whereas the characteristic of a right aortic arch is that the aorta arches to the right Inasmuch as this variant occurs in approximately 25 per cent of all patients with a tetralogy of Fallot, the possibility should always be investigated (see Chapter XXVI) When the aorta arches to the right, the heart is usually long and narrow and its left border shows a slight concavity or a gentle slope (see Figures VI-19 and 20) The aortic knob is frequently visible to the right of the sternum within the shadow of the superior vena cava Even when the x ray or fluoroscopic examination does not show the aorta on the right, upon delineation of the esophagus by barium-opaque mixture, the esophagus is seen to lie to the left of the great vessels Under such circumstances,

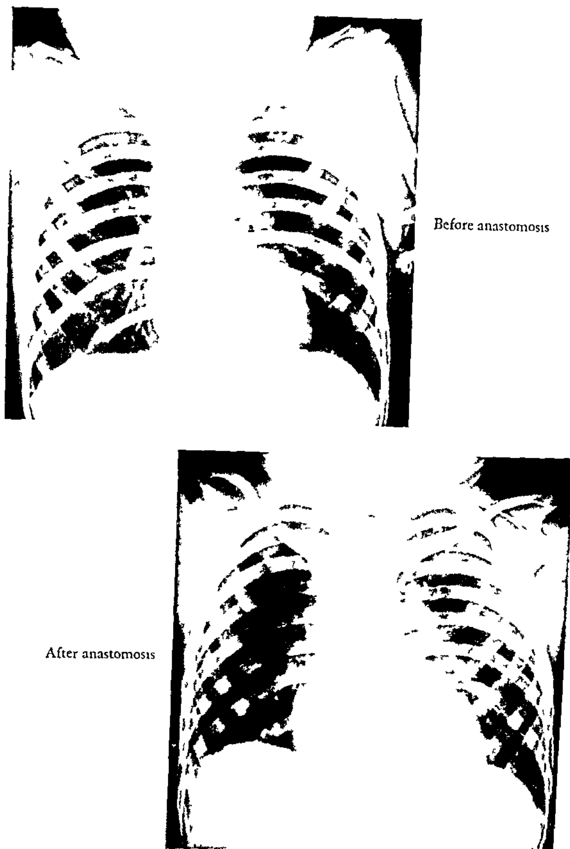
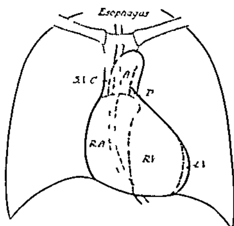
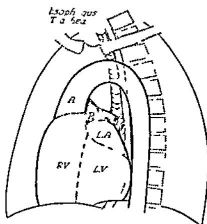


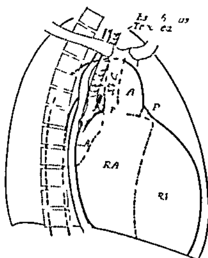
Figure 11–15 Tetralogy of Fallot with a right aortic arch Child



Anterior posterior position



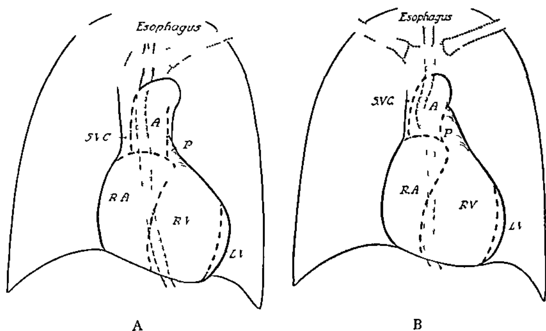
Left anterior-oblique position



Right anterior-oblique position

FIGURE VI-16 Tetralogy of Fallot Child

FIGURE VI-17 Tetralogy of Fallot with a left aortic arch Adult



A

B

FIGURE VI-18 (A) Tetralogy of Fallot with a left aortic arch and (B) normal heart Adult

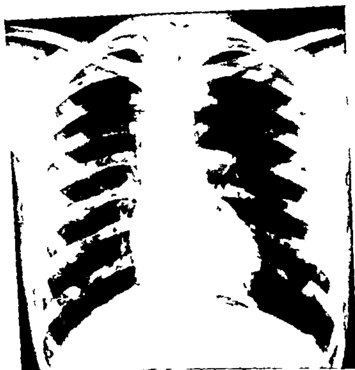


Figure vi-19 Tetralogy of Fallot with a right aortic arch Adult

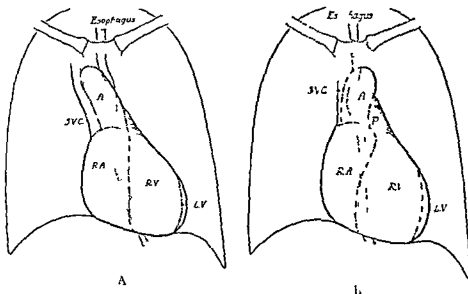


Figure vi-20 (A) Tetralogy of Fallot with a right aortic arch and (B) normal heart Adult

if encroached upon by the aorta, the esophagus is deviated to the left and indented on its right margin by the aorta, as shown in Figures vi–19 and 20 (see also Chapter xxvi)

In the left anterior oblique position the right ventricle is seen to be enlarged, it projects forward toward the anterior chest wall Because of the anterior displacement of the aorta, the aortic shadow lies farther from the spinal column and closer to the anterior chest wall than in the normal heart It requires minimal rotation of the patient for the left ventricle to clear the spinal column The pulmonary window is abnormally clear Furthermore, in cases of a right aortic arch, the relation of the esophagus to the aorta offers additional evidence of the existence of this anomaly *It is in this position, and not in the right anterior* oblique position, that the aorta may be seen to indent the esophagus (see Figure vi–21, LAO position, and also Chapter xxvi)

In the right anterior-oblique position there is no evidence of enlargement of the left auricle Usually there is a striking absence of the fullness of the pulmonary conus (see Figures vi–12 and 13, RAO position) Furthermore, if there is a right aortic arch, the esophagus is seen to descend in a perfectly straight line unrelated to the arch of the aorta (see Figure vi–21, RAO position)

The hilar shadows should be studied with care Owing to the diminished pulmonary circulation, the lungs are usually abnormally clear In older children a tremendous network of small vessels of collateral circulation may develop in the hili of the lungs, these may become so extensive as to cause an increase in the hilar markings If the observer's eyes are fully compensated, these shadows can be seen to be composed of small elements of varying opacity in which ex*pansile pulsations are virtually never visible*

In the evaluation of the hilar shadows it is important to remember that development of collateral circulation and that of polycythemia go hand in hand Therefore, if polycythemia *is slight or absent, increased vascular markings* should never be attributed to collateral circulation

In patients with only slight cyanosis the pulmonary artery is usually relatively large and may occasionally show slight pulsations If, however, the patient is intensely cyanotic, there should be no pulsations in these shadows Furthermore, as the child grows, the heart and great vessels increase in size It follows that the pulmonary artery is more readily visible in older children than in infants Consequently the hilar shadows should always be evaluated in relation to the patient's age and the clinical findings

Slight fullness of the pulmonary conus occurs in a small percentage of pa

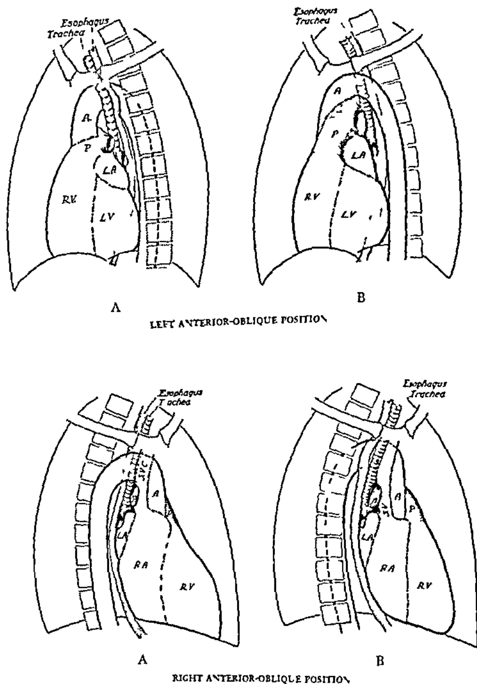


FIGURE VI-21 (A) Tetralogy of Fallot with a right aortic arch and (B) normal heart Adult

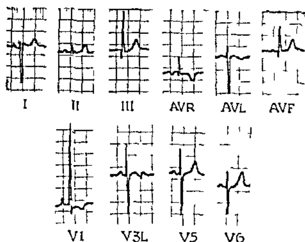


FIGURE VI-22 Tetralogy of Fallot

tients In such instances the pulmonary arteries are readily seen and appear to be of normal size This contour is more frequently encountered in young adults than in infants, partly because the vessels are larger and partly because the pulmonary stenosis is relatively mild Indeed, many of these individuals do so well that medical attention is not sought in infancy and early childhood When the pulmonary artery is relatively large, it raises the question of valvular pulmonary stenosis combined with a fair sized infundibular chamber, or of a valvular pulmonary stenosis with an intact ventricular septum Patients who show such features may require cardiac catheterization, angiocardiography, or both to confirm the diagnosis

ELECTROCARDIOGRAPHIC FINDINGS

A definite right axis deviation in the standard leads and evidence of right ventricular hypertrophy in the unipolar precordial leads are characteristic of this malformation, but there is rarely the prolongation of the intrinsicoid deflection or the deep inversion of T waves in V_1, such as is commonly seen in severe pulmonary stenosis with an intact ventricular septum Absence of right ventricular hypertrophy in the precordial leads is a reason to doubt the diagnosis and is a definite indication for further studies The P waves are frequently abnormally high and pointed (see Figure VI-22) The T waves may or may not be abnormal

SPECIAL TESTS

The circulation time (venous to systemic) is abnormally short In children it is usually between four and six seconds and in adults between eight and twelve seconds The occurrence of a twenty second circulation time should always arouse suspicion that the aorta does not over ride the right ventricle

The oxygen saturation of the arterial blood is abnormally low and falls still lower with exercise A rapid fall in the oxygen saturation of the arterial blood with exercise is characteristic of this malformation When there is doubt as to the presence of cyanosis, exercise will usually render the cyanosis readily apparent

The author had one patient in whom the oxygen saturation of the arterial blood dropped from 83 per cent to 33 per cent on slight exertion, and another in whom it fell from 41 per cent to 15 per cent upon climbing two steps three times When the oxygen saturation of the arterial blood is between 30 per cent and 35 per cent the child can walk only a few feet When the oxygen saturation is between 20 per cent and 25 per cent the child is seldom able to walk Such a child has barely enough oxygen to meet the basal requirements of the body

During infancy the oxygen saturation of the arterial blood may be extraordinarily low It is quite common to find that the arterial oxygen saturation drops to 30 per cent with exercise or vigorous crying If there is a harsh systolic murmur, such a drop in oxygen saturation need cause no undue concern If the oxygen saturation drops to 20 per cent or less, there is reason for concern Most infants lose consciousness when the oxygen saturation drops to 10 per cent or less For such an infant early operation is indicated, especially if the murmur is faint or absent

Effect of exercise on oxygen consumption Usually the pulmonary stenosis is so severe that the patient is unable appreciably to increase the pulmonary blood flow with exercise Consequently exercise causes both a fall in the oxygen saturation and a fall in the oxygen consumption per liter of ventilation [18] This test is however, not diagnostic of a tetralogy of Fallot

Cardiac catheterization may be of aid in the confirmation of the diagnosis, especially when there is fullness of the pulmonary conus or there are slight pulsations in the pulmonary arteries In order to prove the existence of pulmonary stenosis it is essential to catheterize the pulmonary artery and to obtain pressure readings or pressure tracings in the pulmonary artery and in the right ventricle The finding of high pressure in the right ventricle, combined with low pressure in the pulmonary artery, is indicative of pulmonary stenosis In a tetralogy of Fallot, due to the over riding of the aorta, the pressure in the right ventricle usually approaches but seldom exceeds the systemic pressure The author has, however, seen one patient with a tetralogy of Fallot and an extremely small pulmonary artery who ten years after a successful Blalock Taussig operation had developed extreme right ventricular hypertrophy and had a pressure of 250 mm

of mercury in his right ventricle If the catheter enters the aorta, it usually means that the aorta arises in part or entirely from the right ventricle

Although the shunt is primarily from right to left, there is usually a small left to right shunt Therefore it is common to find a slight increase in the oxygen content of the sample of blood taken from the right ventricle as compared with that taken from the right auricle, especially when the sample is taken from the outflow tract of the right ventricle, frequently the oxygen content of the blood in the pulmonary artery is slightly higher than that in the right ventricle This finding is consistent with, but not diagnostic of, a tetralogy of Fallot

Cardiac catheterization can definitely establish the diagnosis of a tetralogy of Fallot, provided that both the aorta and the pulmonary artery are catheterized and that pressure tracings and blood samples are obtained from the various chambers This is, however, seldom possible Furthermore, catheterization gives no clear indication of the degree of dextroposition of the aorta Inasmuch as cardiac catheterization seldom definitively establishes the diagnosis, if the clinical findings are characteristic of a tetralogy of Fallot catheterization is not a prerequisite for operation

Angiocardiography is the most useful test in the demonstration of dextroposition of the aorta When the aorta arises in part or entirely from the right ventricle, it is the rule to see dense filling of the aorta which occurs simultaneously with the filling of the pulmonary artery (see Figure VI–23) This, however, does not always occur The author has studied one infant in whom, although the aorta did not fill until after the dye had circulated through the lungs and returned to the left auricle and left ventricle, operation revealed a fairly large ventricular septal defect and an overriding aorta Furthermore, angiocardiography does not give reliable evidence concerning the size of the pulmonary artery The pulmonary artery may appear to be of moderate size in the angiocardiogram and prove to be extremely small at operation, or the reverse may be true

The aorta is usually better visualized in the anterior posterior view than in the lateral view Therefore, when only a small amount of dye is shunted into the aorta, early delineation of the aorta may be seen in the anterior posterior film and not in the lateral film Nevertheless, lateral films are essential in order to be sure that there is no dye in the left auricle If dye is immediately shunted into the left auricle, there is either an auricular defect or patency of the foramen ovale In either case dye may reach the aorta by way of the left ventricle simultaneously with the filling of the pulmonary artery from the right ventricle Con

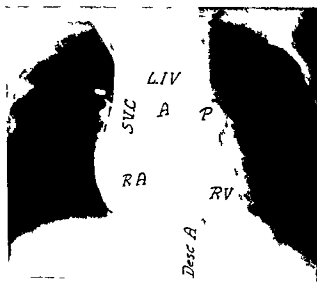




FIGURE VI–23 Tetralogy of Fallot Child

of mercury in his right ventricle If the catheter enters the aorta, it usually means that the aorta arises in part or entirely from the right ventricle

Although the shunt is primarily from right to left, there is usually a small left to-right shunt Therefore it is common to find a slight increase in the oxygen content of the sample of blood taken from the right ventricle as compared with that taken from the right auricle, especially when the sample is taken from the outflow tract of the right ventricle, frequently the oxygen content of the blood in the pulmonary artery is slightly higher than that in the right ventricle This finding is consistent with, but not diagnostic of, a tetralogy of Fallot

Cardiac catheterization can definitely establish the diagnosis of a tetralogy of Fallot, provided that both the aorta and the pulmonary artery are catheterized and that pressure tracings and blood samples are obtained from the various chambers This is, however, seldom possible Furthermore, catheterization gives no clear indication of the degree of dextroposition of the aorta Inasmuch as cardiac catheterization seldom definitively establishes the diagnosis, if the clinical findings are characteristic of a tetralogy of Fallot catheterization is not a prerequisite for operation

Angiocardiography is the most useful test in the demonstration of dextroposition of the aorta When the aorta arises in part or entirely from the right ventricle, it is the rule to see dense filling of the aorta which occurs simultaneously with the filling of the pulmonary artery (see Figure VI–23) This, however, does not always occur The author has studied one infant in whom, although the aorta did not fill until after the dye had circulated through the lungs and returned to the left auricle and left ventricle, operation revealed a fairly large ventricular septal defect and an over riding aorta Furthermore, angiocardiography does not give reliable evidence concerning the size of the pulmonary artery The pulmonary artery may appear to be of moderate size in the angiocardiogram and prove to be extremely small at operation, or the reverse may be true

The aorta is usually better visualized in the anterior posterior view than in the lateral view Therefore, when only a small amount of dye is shunted into the aorta, early delineation of the aorta may be seen in the anterior posterior film and not in the lateral film Nevertheless, lateral films are essential in order to be sure that there is no dye in the left auricle If dye is immediately shunted into the left auricle, there is either an auricular defect or patency of the foramen ovale In either case dye may reach the aorta by way of the left ventricle simultaneously with the filling of the pulmonary artery from the right ventricle Con

trophy As the ductus arteriosus undergoes obliteration, the infant with a tetralogy of Fallot usually develops paroxysmal dyspnea and persistent cyanosis

In an older patient the history is significant in that cyanosis usually dates from infancy and that in childhood squatting was a common habit Physical examination reveals cyanosis and clubbing, in combination with a heart of normal size Usually there is a systolic murmur and a thrill maximal along the left sternal border The second sound at the base is faint and pure The x ray shows a concave curve at the base of the heart to the left of the sternum The pulmonary arteries may or may not be seen The aorta may arch either to the right or to the left. Upon fluoroscopy hilar pulsations are minimal or absent The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

In general, the more severe the pulmonary stenosis, the clearer are the lung fields and the fainter is the murmur In older patients collateral circulation develops and polycythemia increases With intense cyanosis and marked polycythemia, the hilar shadows may become accentuated If however, there is no polycythemia, dense hilar shadows should *not* be attributed to collateral circulation By and large it is rare for an adult with a tetralogy of Fallot to have no *murmur* because it is rare for an individual with a tetralogy of Fallot and a severe pulmonary stenosis to live to adult life *If there* is no murmur and also no great limitation of activity, the patient does not have a tetralogy of Fallot

DIFFERENTIAL DIAGNOSIS

A tetralogy of Fallot must be differentiated from a small ventricular septal defect tricuspid atresia, truncus arteriosus with reduced pulmonary blood flow, and complete transposition of the great vessels with or without pulmonary stenosis, sometimes from a single *ventricle with* pulmonary atresia, or even from pure pulmonary stenosis, and occasionally from primary pulmonary hypertension or defective development of the right ventricle with pulmonary stenosis, or even from the Eisenmenger complex and Ebstein's anomaly of the tricuspid valve

A ventricular septal defect of the maladie de Roger type may require differentiation from a tetralogy of Fallot in early infancy before the appearance of cyanosis A simple ventricular septal defect may, in early infancy, be associated with a slight right axis deviation, *but the precordial leads* seldom show evidence of hypertrophy of either the right or the left ventricle Therefore, the occurrence of right ventricular hypertrophy during the first months of life in an infant with a harsh systolic murmur should arouse suspicion that the condition may prove

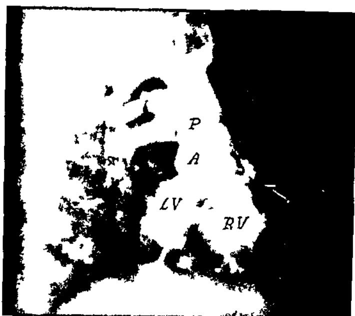


FIGURE VI–24 Tetralogy of Fallot Child

Selective angiocardiogram with dye injected into the outflow tract of the right ventricle

sequently, in the presence of a shunt at the auricular level, it is impossible to prove the dextroposition of the aorta by a *venous* angiocardiogram In such instances, selective angiocardiography with dye injected into the right ventricle eliminates the possibility of a shunt at the auricular level This technique is also best for the visualization of the outflow tract of the right ventricle, as shown in Figure VI–24

DIAGNOSIS

A tetralogy of Fallot with severe pulmonary stenosis or pulmonary atresia is to be suspected in an infant with a heart of normal size and no murmur, who has ceased to gain weight and suffers from severe attacks of paroxysmal dyspnea If the x ray shows a small heart and phenomenally clear lung fields and the electrocardiogram shows both a right axis deviation and evidence of right ventricular hypertrophy, such an infant almost certainly has a tetralogy of Fallot and is in danger of dying from anoxemia

A tetralogy of Fallot with pulmonary stenosis is to be suspected in a young infant with normal color, a heart of normal size, and a harsh systolic murmur, if the electrocardiogram shows evidence of definite right ventricular hyper

Pulmonary stenosis with an intact ventricular septum may occasionally re quire differentiation from a tetralogy of Fallot In early infancy it is the normal ity of the heart in the two conditions which causes the confusion Infants with pure pulmonary stenosis usually gain better than those with a tetralogy of Fallot and they do not suffer from attacks of paroxysmal dyspnea Evidence of progressive cardiac enlargement should always suggest that the pulmonary stenosis is associated with a closed ventricular septum The electrocardiogram frequently shows evidence of right ventricular "strain" in pure pulmonary ste nosis and only rarely does this occur in a tetralogy of Fallot

In older children and young adults the contours of the heart may occasion ally be quite similar in the two conditions A history of squatting, combined with marked limitation of exercise and a sharp fall in the oxygen saturation of the arterial blood upon exercise, is strongly suggestive of a tetralogy of Fallot

In the rare patient who, upon fluoroscopy, does show fullness of the *pulmo*nary conus and slight pulsations in the main branches of *the pulmonary artery*, cardiac catheterization or *angiocardiography may be necessary* in order to de termine whether or not the aorta over rides the right ventricle If, upon cathe *terization, the* catheter enters the aorta, it is proof positive that the aorta arises in part or entirely from the right ventricle On the other hand, if the pressure in the right ventricle is definitely higher than the systemic pressure, that is usually good evidence of an intact ventricular septum Angiocardiography shows that in a tetralogy of Fallot the dye enters the aorta promptly and in a high concen tration whereas in "pure" pulmonary stenosis the dye lingers for a long time in the main pulmonary artery

Primary pulmonary hypertension may be confused with a tetralogy of Fallot during the first months of life Both malformations may cause deep cyanosis Furthermore, the contours of the heart may be closely similar before the pulmonary artery becomes dilated In primary pulmonary hypertension the infant, though cyanotic, does not suffer from *attacks of paroxysmal* dyspnea, the pul monary second sound is usually *accentuated* Over a period of months the heart shows *progressive cardiac enlargement*, which immediately differentiates the condition from a tetralogy *of Fallot* In doubtful cases cardiac catheterization will readily differentiate the *two* conditions, as in a tetralogy of Fallot the pul monary pressure is low, whereas in a primary pulmonary hypertension it is high

The Eisenmenger complex should not be *confused* with a tetralogy of Fallot The late development of cyanosis, the *increased* vascularity of the hilar mark ings, and the presence of a hilar *dance* all differentiate this condition from a tetralogy of Fallot In the less *severe cases* the murmur in a tetralogy of Fallot

to be a tetralogy of Fallot Nevertheless, as long as the infant is doing well and gaining weight there is no cause for concern and no necessity for special studies to determine the exact nature of the malformation

Tricuspid atresia is associated with electrocardiographic evidence of a left axis deviation and of left ventricular hypertrophy This finding readily differentiates it from a tetralogy of Fallot

Truncus arteriosus with markedly reduced pulmonary blood flow may be confused with a tetralogy of Fallot in which there is a severe degree of pulmonary stenosis in early infancy In a truncus arteriosus the heart is usually enlarged and occupies a horizontal position and the pulmonary artery cannot be visualized Cyanosis dates from birth and is intense, polycythemia develops early but in spite of this the infant does better than might be anticipated Such infants rarely suffer from attacks of paroxysmal dyspnea

Complete transposition of the great vessels may occasionally be confused with a tetralogy of Fallot In infants with a tetralogy of Fallot the lungs are remarkably clear, whereas in complete transposition of the great vessels the vascular markings extend nearly to the periphery of the lungs

During infancy, when a complete transposition of the great vessels is combined with a severe pulmonary stenosis, the condition may be extremely difficult to differentiate from a tetralogy of Fallot in which the pulmonary stenosis is severe The quality of the second sound at the base is of prime diagnostic value In a tetralogy of Fallot with pulmonary stenosis, the second sound at the base *to the left of the sternum is faint, whereas in a complete transposition of the* great vessels the aorta lies far to the left and close to the sternum and consequently the second sound in this area is accentuated

Complete transposition of the great vessels may occasionally be compatible with life for a number of years, especially when there is pulmonary stenosis In childhood this condition may be confused with that of a tetralogy of Fallot with extreme pulmonary stenosis Patients with both types of malformations are severely incapacitated and frequently squat when tired Cyanosis and polycythemia are intense In patients with complete transposition of the great vessels, stunting of growth may be extreme The systolic murmur is usually not very loud, but the second 'pulmonic' sound is loud and may be reduplicated The confusion in diagnosis arises because the increased vascular markings are interpreted as due to collateral circulation In a complete transposition of the great vessels the vascular markings extend to the periphery of the lungs, furthermore, they are usually better seen in the x ray film than upon fluoroscopy (see Chapter x)

when one pulmonary artery is well visualized and no dye enters the other pulmonary artery (see Figure VI–25) Needless to say, if an anastomosis is to be performed, it must be performed on the side on which there is a pulmonary artery

Pulmonary insufficiency occurs in the rare instances in which the pulmonary valves are absent Such cases, which are similar to the one mentioned in Chapter II have been reported by Miller et al [1] The pulmonary insufficiency causes an early diastolic murmur which is audible along the left sternal border Such a murmur may also be caused by aortic insufficiency, but this is rare and almost invariably due to acquired disease, therefore it is seldom heard in infancy

A partial anomaly of the venous return may occur Such an anomaly is compensatory in that it increases the oxygen content of the blood in the right ventricle If total correction is undertaken, this abnormality, too, will require correction

Large ventricular septal defects may be associated with mild pulmonary stenosis In such instances there is a large left-to-right shunt and sufficient pulmonary stenosis to break the force with which the blood is ejected to the lungs The increased work required of the right ventricle causes the stenotic area to hypertrophy and consequently the pulmonary stenosis may become progressively greater Over a period of years some infants who suffer from a large left-to-right shunt develop pulmonary stenosis Occasionally the pulmonary stenosis becomes so great that the direction of the shunt is reversed and the patient suffers from reduced pulmonary flow Thus the condition develops into a tetralogy of Fallot Gasul et al have reported a number of such patients who in early infancy suffered from a large left-to-right shunt and who subsequently suffered from reduced pulmonary blood flow When closure of the ventricular septal defect becomes a safe procedure for infants, such infants may be benefited by early correction of the defect (see Chapter XXIV)

COMPLICATIONS

The major complications are due to anoxemia and polycythemia and to the marked increase in the viscosity of the blood, and also to the alteration in its clotting mechanism which develops to compensate for the increased viscosity

Cerebral thrombosis is by far the most common of the serious complications (see Chapter V) Thromboses are liable to occur when polycythemia becomes extreme Multiple minute thromboses are relatively common, large thromboses are not rare The danger of thrombosis is increased by dehydration, therefore, during infections and in hot weather, care should be taken to maintain a high

is always loud, whereas in an Eisenmenger complex it is subject to great variation in intensity The second sound is *never reduplicated in a tetralogy of Fallot*, whereas reduplication of the second sound is common in the Eisenmenger complex Furthermore, in the latter condition cardiac catheterization shows high pressure in the pulmonary artery as well as in the right ventricle

A single ventricle with reduced pulmonary blood flow may be confused with a tetralogy of Fallot Usually either the contour of the heart or the electrocardiogram suggests that the condition is not that of a tetralogy of Fallot Since the rudimentary chamber occupies the position of the outflow tract of the right ventricle, the contour becomes more nearly square than is common in a tetralogy of Fallot The electrocardiogram may show a right axis deviation, but generally does not show the usual pattern of right ventricular hypertrophy in the precordial leads Often there is a deep S wave in all six precordial leads

Defective development of the right ventricle with pulmonary stenosis and an intact ventricular septum may closely simulate a tetralogy of Fallot The absence of paroxysmal dyspnea and failure to squat differentiate this malformation from a tetralogy of Fallot Furthermore, the electrocardiogram frequently shows evidence of the transition from right to left ventricular preponderance in V_2 or V_3 (see Chapter IX, Section A)

Ebstein's anomaly of the tricuspid valve may occasionally be confused with a tetralogy of Fallot In the former condition the infant does not suffer from attacks of paroxysmal dyspnea The heart may be greatly enlarged, and the liver is often engorged but no pulsations are palpable at its margin In infants the electrocardiographic evidence of low voltage in V_1, which is so characteristic of Ebstein's anomaly, is seldom seen In older children, however, the electrocardiogram is frequently of diagnostic aid

COMMONLY ASSOCIATED MALFORMATIONS

A gross defect in the auricular septum is probably the commonest additional malformation This combination of abnormalities is sometimes spoken of as a *pentalogy of Fallot* The condition usually causes no difficulty and is detected only by cardiac catheterization Recognition of the auricular defect is, however, of importance in the surgical treatment of a tetralogy of Fallot (see below)

A single pulmonary artery occasionally occurs in association with a tetralogy of Fallot[19] Nadas et al[20] have also reported the occurrence of unilateral pulmonary atresia The difference in the vascular shadows in the x ray film offers the clue to the diagnosis This diagnosis is readily confirmed by angiocardiography

fluid intake This is especially important in patients with a tetralogy of Fallot, as the malformation is amenable to surgery and consequently compatible with relative longevity For this reason an assiduous effort should be made to prevent cerebral thrombosis and the permanent handicap which results from a hemi plegia

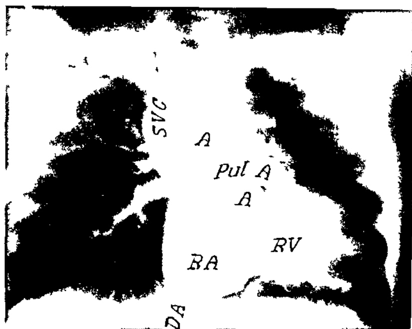
Brain abscess is also a very serious complication Persons in whom venous blood is shunted directly into the systemic circulation are especially prone to such an infection The liability to abscess formation is increased by the minute thrombi and consequent injury to the brain which occurs with long standing polycythemia The differentiation between a brain abscess and cerebral throm bosis is extremely important (see Chapter v) Before the age of two cerebral thromboses are common and brain abscesses are rare Generally the onset is in sidious, the symptoms are progressive, and the patient runs a low grade fever If there is any possibility that a brain abscess exists, an immediate effort should be made to establish or exclude the diagnosis, as prompt treatment is essential

Subacute bacterial endocarditis although frequently curable, is an extremely serious complication Therefore the patient should receive the usual preventive and therapeutic treatment (see Chapter v) If there is any suspicion of a hidden infection, operation for the amelioration or correction of a tetralogy of Fallot should never be undertaken until the danger of infection has been *eliminated* This is an extremely important precaution, because a fresh-cut surface or an anastomosis renders these patients extremely susceptible to subacute bacterial endocarditis

Fever may be caused by severe anoxemia for this reason if a patient has se vere anoxemia and no demonstrable infection, operation should not be post poned because of a persistent low grade fever, as such a patient is in danger of dying from anoxemia

Hemoptysis is a rare complication in patients with a tetralogy of Fallot In most instances the reduction in the pulmonary blood flow is so great that hemop tyses seldom occur Nevertheless, some patients with extensive collateral circula tion do develop hemoptyses, these are due in part to the great strain placed upon the small arterial vessels which lead to the lungs and in part to the bleeding tendency which develops when polycythemia becomes excessive (see Chapter iv)

Hematemesis or *intestinal bleeding* may also occur in a patient with long standing polycythemia especially when, as in a tetralogy of Fallot, the condi



Auricular diastole and ventricular systole



Auricular systole and ventricular diastole

FIGURE VI-25 Tetralogy of Fallot with a single pulmonary artery Child

and the hematocrit reading back to approximately normal values and overcomes these dangers

Surgical treatment is of great benefit to patients with a tetralogy of Fallot Inasmuch as their incapacity is primarily due to insufficient pulmonary blood flow combined with a persistent venous-arterial shunt, any operation which increases the circulation to the lungs ameliorates the condition A Blalock Taussig operation, [3] namely, the anastomosis of a systemic artery to a branch of the pulmonary artery, gives excellent results The same is true of a Potts anastomosis, [4] that is, an anastomosis between the descending aorta and the pulmonary artery It is also possible to insert a graft between the aorta and the pulmonary artery Sir Russell Brock[7] advocates the excision of the stenotic area Lillehei [8] recommends closure of the ventricular septal defect and excision of the stenotic area under direct vision Each operation has its merits and its limitations

The Blalock Taussig operation places remarkably little strain on the heart Immediately after operation cardiac enlargement occurs to adjust to the altered circulation and the increased demands placed upon the heart Thereafter there is no further cardiac enlargement Indeed, if the anastomosis fails to grow with the child, the load on the heart is lessened and it, too, fails to grow with the child, consequently the heart again becomes relatively small

The standard operation for children is the anastomosis of the proximal end of the subclavian artery which arises from the innominate artery to the side of the right or left pulmonary artery Hence the operation is performed on the opposite side to the arch of the aorta The end to-side anastomosis permits the flow of blood to both lungs After operation a good continuous murmur should be audible over both lungs posteriorly The course of the circulation after operation is shown in Diagram VI-4

The mortality rate in the last one hundred patients operated upon between two and twelve years of age was 2 per cent, hence it is a remarkably safe operation for young children

The ideal time for operation is between eight and twelve years of age Only 5 per cent of the patients in this age group have required a second operation within ten years, whereas among younger patients, that is, between four and eight years of age, 32 per cent have required a second operation within ten years but only 4 per cent have had a second operation within five years This means that there is at least a 75 per cent chance that a child in this age group will maintain the benefit derived from operation until he has attained sufficient growth so that total correction will give permanent benefit (see below)

tion is associated with a large systemic blood flow and a small pulmonary blood flow

Hemorrhagic diatheses develop in patients with long standing polycythemia Such alteration in the blood plasma increases the risk of operation and therefore, whenever possible, operation should be performed before such changes have been of long duration In the author s experience, even though alteration in the clotting mechanism has been demonstrated in young individuals, postoperative bleeding has not been a serious complication in patients under twelve years of age

TREATMENT

Medical and surgical treatment are both important

Medical treatment is mainly directed to the relief of paroxysmal dyspnea and to the prevention of complications For a detailed discussion of the following conditions, see Chapter v

An attack of paroxysmal dyspnea calls for prompt treatment The infant should be immediately placed in the knee-chest position or held over the mother's shoulder with his legs doubled up beneath him This simple procedure frequently relieves the attack Oxygen may help to lessen the dyspnea If the attack is long and severe or progresses to loss of consciousness, the infant should be given morphine in full doses, as its action is almost specific The dose is 1 mgm per 4 5 kg (i e, 10 lb) of body weight Within ten to fifteen minutes after receiving morphine intramuscularly the infant usually breathes more easily, his color improves, and the attack gradually wears off

Prophylactic chemotherapy during infections and prior to and following dental extraction or tonsillectomy is extremely important both to prevent subacute bacterial endocarditis and to lessen the danger of brain abscess Intensive chemotherapy is indicated in septicemia, in severe injuries, and also for women during delivery

The author is not yet convinced that prophylactic antibiotics are indicated in an effort to prevent sporadic cases of subacute bacterial endocarditis, in part because if it is to be effective, medication must be given for life, and in part because 100 per cent protection is not humanly possible

Adequate fluid intake in hot weather and during episodes of fever aids in the prevention of dehydration, and thereby lessens the risk of cerebral thrombosis, hemiplegia, and that rare but tragic complication—*blindness* Successful operation brings the red blood cell count, the level of the available hemoglobin,

DIAGRAM VI–4

Tetralogy of Fallot with pulmonary stenosis after closure of the ductus arteriosus and after the Blalock Taussig operation

In a tetralogy of Fallot after the creation of an artificial ductus arteriosus by the anas tomosis of the proximal end of the subclavian artery to the side of the pulmonary ar tery the circulation of the blood is essentially the same as it was before the closure of the ductus arteriosus

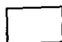
The blood from the right auricle flows into the right ventricle Part of the blood from the right ventricle is pumped out through the stenosed pulmonary artery to the lungs and part is pumped directly into the aorta All the blood in the pulmonary ar teries goes to the lungs and the oxygenated blood is returned by the pulmonary veins to the left auricle and thence it passes to the left ventricle. The blood from the left ventricle is pumped out into the aorta Inasmuch as the aorta over rides the ventricular septum some of the blood from the right ventricle is also pumped directly into the aorta Therefore the aorta and the systemic circulation always receive a mixture of oxygenated blood from the lungs and venous blood from the right ventricle

Most of the blood in the aorta is directed to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle Owing to the pulmonary stenosis and the low pressure in the pulmonary artery some blood from the aorta flows through the artificial ductus arteriosus into the pulmonary artery Thus the lungs receive blood from the right ventricle and also from the aorta through the artificial ductus arteriosus The combined blood flow to the lungs means that a rela tively large volume of blood reaches the lungs All the blood which reaches the lungs is returned by the pulmonary veins to the left auricle and thence to the left ventricle Consequently a relatively large volume of oxygenated blood is pumped out from the left ventricle This blood is mixed with the relatively small volume of blood which is pumped from the right ventricle into the aorta The resultant admixture of venous and arterial blood may be insufficient to produce visible cyanosis

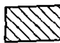
Clinical diagnosis Color is excellent The lips are of normal color The fingers may show slight cyanosis The clubbing recedes The heart may be slightly larger than be fore operation Its contour is essentially the same The systolic murmur, audible before operation usually becomes harsher In addition, a loud continuous murmur is heard throughout the chest this continuous murmur is usually better heard posteriorly than anteriorly

Diagram VI–4

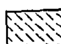
Subclavian Artery anastomosed to Right Pul Art
Head and upper extremities
R Lung
L Lung
P.A.
Pul. A.
PV
Pul V
Aorta
Pulmonary Artery
S.V.C.
L A
R.A.
LV
R.V.
Descending Aorta
I.V.C.
Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

be sufficiently small to protect the lungs from the high pressure in the right ventricle which is secondary to the over riding of the aorta In the rare instance in which a tetralogy of Fallot has only a valvular pulmonary stenosis, care must be taken not to convert the malformation into an Eisenmenger complex Such an operation may be fatal, as the patient lacks the protective pulmonary hypertension characteristic of an Eisenmenger complex Indeed, unless the septal defect is closed, excision of the stenotic area is not advisable

Corrective surgery is now possible Lillehei[26] was the first to demonstrate that with the aid of a pump and oxygenator it was possible to excise the stenotic area and close the ventricular defect and thereby to restore the heart and circulation to normal Such an operation is probably the ideal for adults and for children who have nearly attained their growth

For infants and young children with severe pulmonary stenosis or pulmonary atresia, in whom the pulmonary artery is but one third to one half as large as the aorta, it seems doubtful that the right ventricle, the pulmonary orifice, and the pulmonary artery will expand to normal size and grow normally with the child When a patch is necessary, it must be made of the proper size for the patient It is manifestly impossible to insert the patch of suitable size for an adult into a tiny infant, hence there is real danger that total correction in infancy or early childhood may leave the individual with an underdeveloped right ventricle and pulmonary stenosis by the time he reaches adolescence

Furthermore, recent experience has shown that a previous Blalock anastomosis does not greatly increase the risk of operation for total correction Moreover, Ferencz [7] studies on the pulmonary vascular bed have shown that, whereas most infants with anoxemia and decreased pulmonary blood flow have multiple thrombi in their lungs, patients with an adequate anastomosis who have died of unrelated causes have a normal pulmonary vascular bed Therefore the child who has had a previous anastomosis may be better able to adjust to a corrective procedure than the patient who has not had a previous operation

The indications for operation vary with the age of the patient and the severity of the pulmonary stenosis A baby with a tetralogy of Fallot who has a small heart but no murmur, and excessively clear lung fields, may be in real danger of dying from anoxemia, even though he has no polycythemia and but slight cyanosis Such infants frequently suffer from repeated attacks of paroxysmal dyspnea If the infant ceases to gain weight and the attacks of paroxysmal dyspnea become long and severe or progress to loss of consciousness, early operation is urgently indicated If, however, the infant is gaining weight and doing well, it

In infants the problem is more difficult The subclavian artery is often too small to be of great benefit More than 33 per cent of patients operated on at the Johns Hopkins Hospital who were under two years of age have required a second operation within ten years Such infants require a relatively large vessel *The sacrifice of the innominate artery has, however, proved too dangerous to the cerebral circulation to justify its use in an effort to obtain a larger vessel* When a large opening is desired, a Potts procedure is preferable to a Blalock Taussig anastomosis

Potts' anastomosis, that is, an anastomosis between the descending aorta and the pulmonary artery, gives excellent results Potts has emphasized the importance of the correct size of the opening He recommends that the length of the incision be exactly 4 mm regardless of the size of the patient Any opening greater than 4 mm in diameter may cause cardiac failure, one of lesser diameter may thrombose Nevertheless, slight variation in the size of the anastomosis is almost inevitable Consequently it requires great experience to obtain uniform results Obviously the operation must be performed on the side of the descending aorta Therefore the operation is difficult in a patient with a right aortic arch *It appears that an anastomosis between the aorta and the pulmonary artery* grows better than a Blalock-Taussig anastomosis Indeed, in a number of instances the diameter of the opening has increased in size This is probably because greater stress and strain are placed on the aorta, as it grows, than on the pulmonary artery Hence there is real danger that the anastomosis may become too large and transmit the high pressure in the aorta to the pulmonary artery and consequently injure the pulmonary vascular bed

The insertion of a graft as formerly advocated by Gross in this country and commonly done in the U S S R , eliminates the sacrifice of the subclavian artery This technique, however, doubles the length of the operation and increases the risk of operation *Although notching of the ribs on the side on which the subclavian artery is sacrificed is relatively common, this does no harm* To the best of the author's knowledge, only one patient in 2,000 has experienced any serious complications from the ligation of the subclavian artery, therefore the insertion of a graft is indicated only in the rare instances when no vessel is obtainable

Direct attack and the excision of the stenotic area, as advocated by Brock, is possible only if a tetralogy of Fallot is associated with a valvular pulmonary stenosis or if there is a fair-sized infundibular chamber *If there is an infundibular stenosis,* a sufficient amount of muscle must be removed so as to prevent subsequent constriction from scar formation and, at the same time, the opening must

be sufficiently small to protect the lungs from the high pressure in the right ventricle which is secondary to the over riding of the aorta In the rare instance in which a tetralogy of Fallot has only a valvular pulmonary stenosis, care must be taken not to convert the malformation into an Eisenmenger complex Such an operation may be fatal, as the patient lacks the protective pulmonary hypertension characteristic of an Eisenmenger complex Indeed, unless the septal defect is closed, excision of the stenotic area is not advisable

Corrective surgery is now possible Lillehei[6] was the first to demonstrate that with the aid of a pump and oxygenator it was possible to excise the stenotic area and close the ventricular defect and thereby to restore the heart and circulation to normal Such an operation is probably the ideal for adults and for children who have nearly attained their growth

For infants and young children with severe pulmonary stenosis or pulmonary atresia, in whom the pulmonary artery is but one third to one half as large as the aorta, it seems doubtful that the right ventricle, the pulmonary orifice, and the pulmonary artery will expand to normal size and grow normally with the child When a patch is necessary, it must be made of the proper size for the patient It is manifestly impossible to insert the patch of suitable size for an adult into a tiny infant, hence there is real danger that total correction in infancy or early childhood may leave the individual with an underdeveloped right ventricle and pulmonary stenosis by the time he reaches adolescence

Furthermore, recent experience has shown that a previous Blalock anastomosis does not greatly increase the risk of operation for total correction Moreover, Ferencz studies on the pulmonary vascular bed have shown that, whereas most infants with anoxemia and decreased pulmonary blood flow have multiple thrombi in their lungs, patients with an adequate anastomosis who have died of unrelated causes have a normal pulmonary vascular bed Therefore the child who has had a previous anastomosis may be better able to adjust to a corrective procedure than the patient who has not had a previous operation

The indications for operation vary with the age of the patient and the severity of the pulmonary stenosis A baby with a tetralogy of Fallot who has a small heart but no murmur, and excessively clear lung fields, may be in real danger of dying from anoxemia, even though he has no polycythemia and but slight cyanosis Such infants frequently suffer from repeated attacks of paroxysmal dyspnea If the infant ceases to gain weight and the attacks of paroxysmal dyspnea become long and severe or progress to loss of consciousness, early operation is urgently indicated If, however, the infant is gaining weight and doing well, it

is wise to postpone operation, as the risk of operation is less and the results are better in childhood than in infancy

The less severe the pulmonary stenosis, the louder is the systolic murmur Therefore, if the infant has a loud systolic murmur, early operation is seldom necessary Indeed, if the attacks of paroxysmal dyspnea can be promptly relieved by placing the infant on his abdomen in the knee chest position, it is usually safe to postpone operation Such attacks are most severe as the ductus arteriosus undergoes obliteration and prior to the development of polycythemia

In brief, the decision concerning operation must be influenced by the frequency and severity of these attacks and by whether the patient is losing ground or improving The determination of the arterial oxygen saturation may be of aid If it is only 30 per cent when the infant is crying, there is no cause for concern If it is under 20 per cent, the condition is serious, and under 10 per cent is dangerously low

The persistence of attacks of paroxysmal dyspnea or loss of consciousness in childhood is an indication for operation If the child s exercise tolerance is extremely limited, that too is an indication for operation The early development of marked polycythemia is also an indication for operation, as when the hematocrit reading reaches 70 per cent there is a real danger of thrombosis

Most parents are anxious for operation before the child starts school If, however, cyanosis is minimal and the red blood cell count is only 6 or 7 million cells per cu mm , and the limitation of exercise is slight, it may be wise to postpone surgery with the hope that only one operation will be necessary

If, however, the child is seriously incapacitated, it is wiser to operate while the child is young and thus give him a normal childhood than to try to postpone operation until total correction is indicated It is always advisable to operate while the patient still has good tissue turgor and before the secondary changes of the lungs due to long standing polycythemia become a serious problem The hemorrhagic diathesis is far more serious in adults than in children Even though a child may have a great reduction in platelets and a low blood fibrinogen, he seldom suffers from severe hemorrhage after the operation, whereas in the young adult hemorrhage in the early postoperative period may be a serious, even fatal, complication Therefore in 1960 it seems to the author advisable to perform a Blalock Taussig operation on an infant or young child and to undertake total correction on older children with mild pulmonary stenosis or young adults who have attained their growth

The occurrence of a gross defect in the auricular septum may increase the strain on the circulation after an anastomotic procedure A number of patients

have developed considerable cardiac enlargement and a few have suffered from cardiac failure Nevertheless, a number of patients in whom cardiac catheterization indicated the presence of an auricular septal defect have had a smooth postoperative course and subsequently have experienced no difficulty from the additional defect Inasmuch as at the Johns Hopkins Hospital cardiac catheterization is not a routine procedure prior to operation, there are no figures available as to the incidence of auricular septal defects in our series of 1,500 patients with a tetralogy of Fallot

The occurrence of an auricular defect does, however, lessen the chance of an excellent result Therefore, when such an anomaly is known to exist, if possible it may be wise to postpone operation until such time as total correction may be of permanent benefit

PROGNOSIS

The prognosis varies with the severity of the pulmonary stenosis When the pulmonary stenosis is extreme or there is pulmonary atresia, the prognosis without operation is extremely poor In most instances closure of the ductus arteriosus renders the condition incompatible with life Therefore early operation is indicated

The tetralogy of Fallot has long been considered as the commonest malformation of the cyanotic group to be compatible with relative longevity Formerly all those who suffered from this malformation were able to receive great encouragement from a case reported by White and Sprague[3] of a man with a tetralogy of Fallot who lived for sixty years and was a concert violinist This man must have had a relatively mild pulmonary stenosis Few persons are as fortunate as he Today, however, persons with a pulmonary stenosis of such severity that it endangers or handicaps their lives can be greatly helped by surgery A Blalock Taussig operation gives a child a 75 per cent chance to enjoy a virtually normal life, at least, until adolescence Indeed, several women who have had this operation have borne normal children Once the patient has attained his growth, there is every reason to believe that total correction of the malformation not only will restore the circulation to normal but will be of permanent benefit Thus the prognosis is greatly improved

SUMMARY

The first known case of a tetralogy of Fallot was reported in 1671, but it was not until 1888 that Fallot clarified the syndrome which now bears his name

The four features which constitute the tetralogy of Fallot are pulmonary ste

nosis, dextroposition of the aorta, a high ventricular septal defect, and right ventricular hypertrophy

The pulmonary stenosis renders it difficult for the blood from the right ventricle to be pumped out to the lungs The dextroposition of the aorta means that some of the blood from the right ventricle which would normally go to the lungs for oxygenation is pumped into the aorta This leads to a reduction in the pulmonary blood flow and an increase in the systemic blood flow The pressure in the right ventricle is approximately the same as that in the systemic circulation The pressure in the pulmonary artery is low The work of the right ventricle is increased, there is right ventricular hypertrophy

The outstanding clinical features are cyanosis, dyspnea, a heart of normal size with a *systolic murmur and a weak pulmonic second sound Cyanosis usually* appears between three and six months of age The intensity of the cyanosis varies not only with the amount of reduced hemoglobin in the circulating blood but also with the amount of available hemoglobin

Cyanosis may be absent at birth The infant gains slowly and frequently suffers from attacks of paroxysmal dyspnea Cessation of weight gain and attacks of dyspnea progressing to loss of consciousness are ominous signs The infant is in danger of dying from anoxemia even though the heart is normal in size and there is no murmur

Cyanosis usually appears during the first year of life Compensatory polycythemia *develops as the collateral circulation becomes established*

Polycythemia gradually increases and may become extreme With long standing polycythemia the blood platelets and the blood fibrinogen become reduced

There is usually cyanosis and clubbing of the extremities The child frequently squats when tired

The heart is normal in size The intensity of the systolic murmur is inversely proportional to the severity of the pulmonary stenosis The pulmonic second sound is reduced in intensity but is usually audible

The x ray shows a small heart with a concave curve at its base to the left of the sternum and decreased hilar markings A right aortic arch occurs in 25 per cent of these patients

The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

The circulation time is short

The oxygen saturation of the arterial blood is low and *falls still further with* exercise

Cardiac catheterization shows that the pressure in the right ventricle approaches systemic pressure and that the pressure in the pulmonary artery is low There is evidence of both a left to-right and a right to-left shunt

Angiocardiography may aid in the demonstration of the dextroposition of the aorta

Selective angiocardiography is of great value in the visualization of the outflow tract of the right ventricle

Diagnosis is based on the size and the contour of the heart and on the electrocardiographic findings of a right axis deviation and right ventricular hypertrophy in a cyanotic child who gives a history of paroxysmal dyspnea and who squats when tired

The condition requires differentiation from other malformations with pulmonary stenosis and a venous arterial shunt namely, pulmonary stenosis with an intact ventricular septum and a patent foramen ovale, a single ventricle, tricuspid atresia, a truncus arteriosus, complete transposition of the great vessels, occasionally from primary pulmonary hypertension or an Eisenmenger complex, and rarely from Ebstein s anomaly of the tricuspid valve

The most common associated anomaly is a gross defect in the auricular septum When this occurs the condition is known as a pentalogy of Fallot Occasionally there is but a single pulmonary artery and even more rarely the pulmonary valves are so abnormal that there is pulmonary insufficiency

The common complications are cerebral thrombosis, brain abscess, and subacute bacterial endocarditis Hemoptyses and hematemeses are rare complications, but do occur

Medical treatment is directed to the relief of paroxysmal dyspnea, to the prevention of infection and to the prevention of thrombosis For the last reason, prior to operation high fluid intake is important

Surgical treatment is of great benefit Any operation which increases the circulation to the lungs without raising the pressure with which the blood is ejected into the lungs is of great help A Blalock Taussig operation gives excellent results in childhood The ideal age for operation is from eight to twelve years, 90 per cent of the children in this age group who obtained good results from operation have maintained their benefi for the ensuing ten years

Closure of the ventricular defect and relief of the pulmonary stenosis by means of open heart surgery restores the circulation to normal The operation is performed with remarkable success in older individuals Not only is it more difficult in young children, but also there is real danger that, if the pulmonary orifice and pulmonary artery are abnormally small, the right side of the heart

nosis, dextroposition of the aorta, a high ventricular septal defect, and right ventricular hypertrophy

The pulmonary stenosis renders it difficult for the blood from the right ventricle to be pumped out to the lungs The dextroposition of the aorta means that some of the blood from the right ventricle which would normally go to the lungs for oxygenation is pumped into the aorta This leads to a reduction in the pulmonary blood flow and an increase in the systemic blood flow The pressure in the right ventricle is approximately the same as that in the systemic circulation The pressure in the pulmonary artery is low The work of the right ventricle is increased, there is right ventricular hypertrophy

The outstanding clinical features are cyanosis, dyspnea, a heart of normal size with *a systolic murmur* and a *weak pulmonic second sound* *Cyanosis* usually appears between three and six months of age The intensity of the cyanosis varies not only with the amount of reduced hemoglobin in the circulating blood but also with the amount of available hemoglobin

Cyanosis may be absent at birth The infant gains slowly and frequently suffers from attacks of paroxysmal dyspnea Cessation of weight gain and attacks of dyspnea progressing to loss of consciousness are ominous signs The infant is in danger of dying from anoxemia even though the heart is normal in size and there is no murmur

Cyanosis usually appears during the first year of life Compensatory polycythemia develops as the collateral circulation becomes established

Polycythemia gradually increases and may become extreme With long standing polycythemia the blood platelets and the blood fibrinogen become reduced

There is usually cyanosis and clubbing of the extremities The child frequently squats when tired

The heart is normal in size The intensity of the systolic murmur is inversely proportional to the severity of the pulmonary stenosis The pulmonic second sound is reduced in intensity but is usually audible

The x ray shows a small heart with a concave curve at its base to the left of the sternum and decreased hilar markings A right aortic arch occurs in 25 per cent of these patients

The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

The circulation time is short

The oxygen saturation of the arterial blood is low and falls still further with exercise

and a living patient fifty-eight years old with cyanosis and clubbing of the fingers Am Heart J 3 77–90 1927

18 Bing R. J., L D Vandam and F D Gray Jr Physiological studies in congenital heart disease results of preoperative studies in patients with tetralogy of Fallot Bull Johns Hopkins Hosp 80 121–141, 1947

19 Blalock A Surgical procedures employed and anatomical variations encountered in the treatment of congenital pulmonic stenosis Surg, Gynec & Obst 87 385–409 1948

20 Nadas A S H D Rosenbaum M H Wittenborg and A M Rudolph Tetralogy of Fallot with unilateral pulmonary atresia clinically diagnosable and surgically significant variable Circulation 8 328–336 1953

21 Miller R A H White and M Lev Congenital absence of the pulmonary valve clinical and pathological syndrome Circulation 18 759 1958 (Abstracts of Thirty first Scientific Sessions of the American Heart Association San Francisco October, 1958)

22 Gasul B M J B Richmond and C A Krakower A case of tetralogy of Fallot with a patent foramen ovale (pentalogy) showing a marked left ventricular hypertrophy and left axis deviation J Pediat 35 413–421 1949

23 Blalock A and H B Taussig The surgical treatment of malformations of the heart in which there is pulmonary stenosis or pulmonary atresia J.A.M.A 128 189–202 1945

24 Potts W J S Smith and S Gibson Anastomosis of the aorta to a pulmonary artery J.A.M.A 132 627–631, 1946

25 Brock R C Pulmonary valvulotomy for the relief of congenital pulmonary stenosis report of 3 cases Brit M J 1 1121–1126 1948

26 Lillehei C W M Cohen H E Warden R C Read J B Aust R A DeWall and R L Varco Direct vision intracardiac surgical correction of the tetralogy of Fallot pentalogy of Fallot and pulmonary atresia defects, report of first 10 cases Ann Surg 14 418–445 1955

27 Ferencz C The pulmonary vascular bed in the tetralogy of Fallot changes following a systemic pulmonary arterial anastomosis Bull Johns Hopkins Hosp 106 100–118, 1960

28 Taussig H B H Crawford S Pelargonio and S Zacharioudakis Ten to thirteen year follow up on 244 patients operated on for pulmonary stenosis and atresia between 1944 and 1940 Reported at the Ninth International Pediatric Congress Montreal 1959 (In press)

will not grow in proportion to the growth of the child Were this to happen, the patient would be left with a pulmonary stenosis and an intact ventricular septum Therefore it seems wise to the author to reserve total correction until the patient has nearly attained his growth

References

1 Stensen, Nils (1638–86) Quoted by H I Goldstein in a discussion of Saul Jarcho s paper *Giovanni Battista Morgagni*, *Bull Hist Med* 20 526, 1948

2 Sandifort s Observationes *See* L R Bennett

3 Bennett L R Sandifort s Observationes, Chapter 1, Concerning a very rare disease of the heart, 1 tetralogy of Fallot or Sandifort? Bull Hist Med 20 539–570, 1946

4 Peacock, T B *On Malformations of the Human Heart, etc, with Original Cases and* Illustrations (2d ed) London, Churchill, 1866 No illustrations of the tetralogy of Fallot were shown in the first edition (published in London in 1858)

5 Fallot, E Contribution a l anatomie pathologique de la maladie bleue (cyanose cardiaque) Marseille med 25 77–93, 138–158, 207–223, 270–286, 341–354, 403–420, 1888

6 Spitzer, A Ueber den Bauplan des normalen und missbildeten Herzens, Versuch einer phylogenetischen Theorie Virchows Arch f path Anat 243 81–272, 1923

7 Keith, A The Hunterian Lectures on malformations of the heart Lancet 2 359–363 433–435 519–523 1909

8 Keith, Sir A K Human Embryology and Morphology (5th ed) London Edward Arnold, 1933

9 Johns T N P, G R Williams, and A Blalock The anatomy of pulmonary stenosis and atresia with comments on surgical therapy Surgery 33 161–172, 1953

10 Hartman, R C A hemorrhagic disorder occurring in patients with cyanotic congenital heart disease Bull Johns Hopkins Hosp 91 49–67, 1952

11 Assmann H Die klinische Roentgendiagnostik der inneren Erkrankungen (4th ed) Leipzig Vogel, 1929

12 White P D and H B Sprague The tetralogy of Fallot, report of a case in a noted musician who lived to his sixtieth year J A M A 92 787–791, 1929

13 Blackford, L M Tetralogy of Fallot clinical report of a case Arch Int Med 45 631–640, 1930

14 Harrison, W F Congenital heart disease, extreme congenital pulmonary stenosis (*tetralogy of Fallot*), *collateral pulmonary circulation, massive right sided vegetative* endocarditis Am Heart J 5 213–231, 1929

15 Papp, C Le diagnostic radiologique de la tetrade de Fallot Arch d mal du coeur 24 249–258, 1931

16 Segall, H N A case of tetralogy of Fallot, clinicopathological observations, quantitative studies of circulation rate and the right to left shunt Am Heart J 8 628–649, 1933

17 Kurtz, C M, H B Sprague, and P D White Congenital heart disease, interventricular septal defects with associated anomalies in a series of 3 cases examined post mortem,



Courtesy of Dr J[illegible] Ed[illegible] and the Mayo Cl[illegible]

FIGURE VII–1 Origin of both great vessels from the right ventricle and both of normal size

also arises from the right ventricle The pulmonary orifice is usually stenosed and the pulmonary valve may be bicuspid The left ventricle lacks its normal outflow tract the only way for blood to leave the left ventricle is through a ventricular septal defect Hence a ventricular septal defect is an integral part of the malformation Indeed, the size of the ventricular defect determines the ease with which blood can be pumped out of the left ventricle and hence the amount of work required of that chamber

The origin of the aorta from the right ventricle makes it the systemic ventricle, hence there is right ventricular hypertrophy but the pulmonary stenosis protects the lungs Unless the ventricular defect is large, there is difficulty in the expulsion of blood from the left ventricle, hence it, *too*, undergoes dilatation and hypertrophy The foramen ovale is closed and the ductus arteriosus undergoes normal obliteration Figure VII–2 illustrates this type of malformation

COURSE OF THE CIRCULATION

During fetal life the course of the circulation is grossly altered by the abnormal position of the aorta Most of the blood from the right auricle which

CHAPTER VII

ORIGIN OF BOTH GREAT VESSELS FROM THE RIGHT VENTRICLE

T*HIS* malformation represents a further rotation of the aorta than that which occurs in the tetralogy of Fallot. The aorta is so far dextroposed that it arises entirely from the right ventricle. The pulmonary artery is usually stenotic and in most instances it occupies its normal position, as it does in the tetralogy of Fallot; thus it, too, arises from the right ventricle. Between 1948 and 1959 we have studied five cases in which both great vessels arose from the right ventricle.[1] Occasionally the stenosed pulmonary artery lies posterior and *slightly to the right of the aorta but arises entirely from the right ventricle.* Under such circumstances the relation of the two great vessels is that of a corrected transposition; nevertheless, both great vessels arise from the right ventricle. This malformation was known to Maude Abbott.[2] Lev and Saphir,[3] in a report of six cases of transposition of the arterial trunk, described three such cases. Witham[4] in 1957 reported the clinical and pathological findings in four cases and reviewed the literature.

If the pulmonary artery is atretic, it makes no difference whether it lies anterior or posterior to the aorta. When an atretic pulmonary artery arises from the left ventricle, the condition constitutes a transposition of the great vessels; therefore the condition is discussed in Chapter x, Section c.

This malformation may also occur with a pulmonary artery of normal size (see Figure VII-1). *Under such circumstances there is adequate circulation to the* lungs, but pulmonary congestion usually occurs early, as a large volume of blood is ejected to the lungs under systemic pressure and great difficulty is encountered in the expulsion of blood from the left ventricle. When the ventricular septal defect is relatively large, the condition may be compatible with life for a number of years. Under such circumstances the malformation *simulates the Eisenmenger* complex but differs from it in that cyanosis dates from birth and the heart undergoes slow progressive enlargement.

The present discussion is limited to the malformation as it occurs with pulmonary stenosis.

NATURE OF THE MALFORMATION

The aorta arises entirely from the right ventricle and the pulmonary artery



Courtesy [illegible] Dr J [illegible] Edward[illegible] the Mayo Cl[illegible]

FIGURE VII-1 Origin of both great vessels from the right ventricle and both of normal size

also arises from the right ventricle The pulmonary orifice is usually stenosed and the pulmonary valve may be bicuspid The left ventricle lacks its normal outflow tract the only way for blood to leave the left ventricle is through a ventricular septal defect Hence a ventricular septal defect is an integral part of the malformation Indeed, the size of the ventricular defect determines the ease with which blood can be pumped out of the left ventricle and hence the amount of work required of that chamber

The origin of the aorta from the right ventricle makes it the systemic ventricle, hence there is right ventricular hypertrophy but the pulmonary stenosis protects the lungs Unless the ventricular defect is large, there is difficulty in the expulsion of blood from the left ventricle, hence it, too, undergoes dilatation and hypertrophy The foramen ovale is closed and the ductus arteriosus undergoes normal obliteration Figure VII-2 illustrates this type of malformation

COURSE OF THE CIRCULATION

During fetal life the course of the circulation is grossly altered by the abnormal position of the aorta Most of the blood from the right auricle which

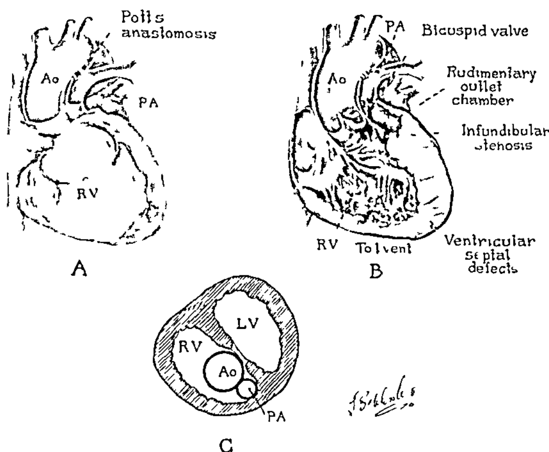


FIGURE VII–2 Origin of both great vessels from the right ventricle and an abnormally small pulmonary artery

flows into the right ventricle is pumped out through the aorta to the body and is returned by the superior vena cava and the inferior vena cava to the right auricle Some of the blood from the right ventricle is pumped out through the pulmonary artery to the lungs, and some blood may flow through the ductus arteriosus to the lungs All the blood which goes to the lungs is returned in the normal manner to the left auricle In addition to the blood returned from the lungs to the left auricle, some blood from the right auricle flows through the foramen ovale to the left auricle All the blood in the left auricle flows into the left ventricle Here difficulty is encountered in the ejection of blood from the left ventricle, because the only way for blood to be pumped out of the left ventricle is through the septal defect to the right ventricle This increases the work of the left ventricle Hence at birth the two ventricles are of approximately equal thickness (see Figure VII–3)

After birth the course of the circulation remains essentially the same The blood from the right auricle flows into the right ventricle and is pumped out into the aorta and also into the pulmonary artery The expansion of the lungs lowers the pulmonary pressure and increases the volume of venous blood which flows through the pulmonary artery and also that which flows from the aorta through the ductus arteriosus to the lungs All the blood which flows to the lungs is oxygenated and returned by the pulmonary veins to the left auricle The increased circulation to the lungs increases the volume of blood returned to the left auricle and tends to close the foramen ovale The blood in the left auricle flows into the left ventricle The increased volume of blood which is returned to the left ventricle increases the volume of blood which must pump through the septal defect to the right ventricle and thence out into the aorta and the pulmonary artery The blood which flows to the body is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle So the cycle continues The course of the circulation is shown in Diagram VII–1

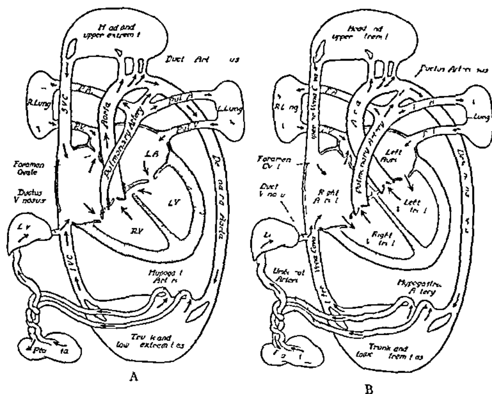


FIGURE VII–3 Fetal circulation (A) Origin of both great vessels from the right ventricle and (B) normal heart

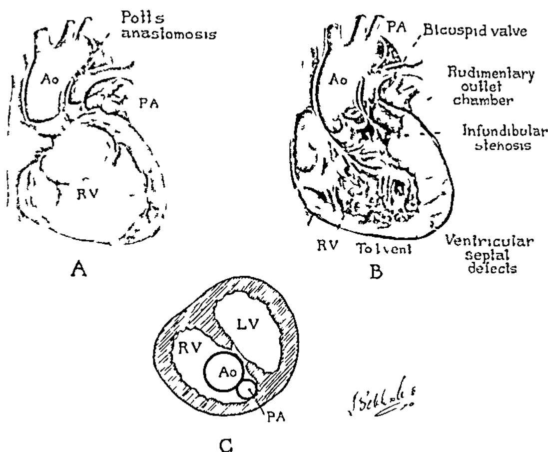


FIGURE VII-2 Origin of both great vessels from the right ventricle and an abnormally small pulmonary artery

flows into the right ventricle is pumped out through the aorta to the body and is returned by the superior vena cava and the inferior vena cava to the right auricle Some of the blood from the right ventricle is pumped out through the pulmonary artery to the lungs, and some blood may flow through the ductus arteriosus to the lungs All the blood which goes to the lungs is returned in the normal manner to the left auricle In addition to the blood returned from the lungs to the left auricle, some blood from the right auricle flows through the foramen ovale to the left auricle All the blood in the left auricle flows into the left ventricle Here difficulty is encountered in the ejection of blood from the left ventricle, because the only way for blood to be pumped out of the left ventricle is through the septal defect to the right ventricle This increases the work of the left ventricle Hence at birth the two ventricles are of approximately equal thickness (see Figure VII-3)

Diagram VII–1

Origin of both great vessels from the right ventricle and a small pulmonary artery

The essential feature of this malformation is that the aorta is transposed and arises from the right ventricle the pulmonary artery is stenosed and *it too, arises from the right ventricle* In order for the blood to escape from the left ventricle there must be a ventricular septal defect.

The blood from the right auricle flows into the right ventricle Most of the blood in the right ventricle is pumped out through the aorta to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle Inasmuch as there is pulmonary stenosis, only a small amount of blood is pumped through the pulmonary artery to the lungs where it is oxygenated and returned to the left auricle So long as the ductus arteriosus remains open, some blood flows through the ductus arteriosus to the lungs This increases the circulation to the lungs and also increases the volume of blood returned to the left auricle and the left ventricle

Since both the aorta and the pulmonary artery arise from the right ventricle the only way for the blood to leave the left ventricle is through the septal defect to the right ventricle If this is small the left ventricle has difficulty in emptying itself hence there is also left ventricular hypertrophy

Clinical diagnosis Inasmuch as the aorta arises from the right ventricle the infant shows persistent cyanosis he does not suffer from attacks of paroxysmal dyspnea but may have episodes of loss of consciousness The heart is enlarged There is absence of fullness of the pulmonary conus Viewed in the left anterior-oblique position the right ventricle is enlarged there may or may not be enlargement of the left ventricle The electrocardiogram shows a right axis deviation and evidence of right ventricular strain Death results from anoxemia or cardiac failure.

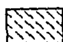
Diagram VII-1



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

X RAY AND FLUOROSCOPIC FINDINGS

The heart is enlarged, especially to the left There is no fullness in the region of the pulmonary conus The cardiac shadow extends downward and outward from a narrow base (see Figure VII-4) The lungs are clear and the hilar markings are inconspicuous

In the left anterior-oblique position both ventricles are seen to be enlarged The right ventricle extends out toward the chest wall and the shadow of the left ventricle extends posteriorly far beyond the spinal column, as seen in Figure VII-5 The small size of the pulmonary artery renders the pulmonary window abnormally clear In addition, the origin of the aorta from the right ventricle causes the pulmonary window to be unusually wide Consequently the pulmonary window is both wide and clear

Examination in the right anterior-oblique position contributes little

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar precordial leads show evidence of right ventricular hypertrophy and the pattern of so-



FIGURE VII-4 Origin of both great vessels from the right ventricle and pulmonary stenosis (same patient as in Figures VII-5 6, 7) Infant

PHYSIOLOGY OF THE MALFORMATION

In this malformation the right ventricle pumps the blood into the aorta, therefore the systolic pressure in the right ventricle is the same as that in the systemic circulation The occurrence of pulmonary stenosis, however, protects the lungs and the pulmonary pressure is low The combined blood flow through the stenosed pulmonary artery and the ductus arteriosus is usually sufficient for life Nevertheless, only a small volume of blood is directed to the lungs and a large volume of mixed venous and arterial blood flows to the body, where it gives up more of its oxygen Hence a small volume of oxygenated blood is mixed with a large volume of venous blood which is extremely low in oxygen content It follows that the oxygen content of the mixed venous and arterial blood is abnormally low

CLINICAL FINDINGS

Cyanosis appears early, it usually dates from birth The infant frequently suffers from "spells" during which the cyanosis increases in intensity The spell may be long and severe and progress to loss of consciousness

Polycythemia develops early and may become extreme The red blood cell count may reach 9 or 10 million cells per cu mm , the level of the available hemoglobin and the hematocrit reading are proportionally high

Dyspnea is usually severe but *attacks of paroxysmal dyspnea* seldom occur

Barrel shaped chest deformity is the rule

The lungs remain clear, as the pulmonary blood flow is meager

Growth and development are severely retarded

Exercise tolerance is markedly restricted Children with this malformation seldom learn to walk before three years of age, thereafter they squat when tired

The liver may be enlarged and pulsations may be detectable at its margin

Edema and ascites are late manifestations of cardiac failure

CARDIAC FINDINGS

The heart is enlarged *A systolic murmur* and a *thrill* are usually present The *second sound* at the base of the heart to the left of the sternum is accentuated owing to the transposed aorta A *gallop rhythm* may be present

Cardiac failure occurs early It is primarily right sided failure with enlargement of the liver and edema of the extremities

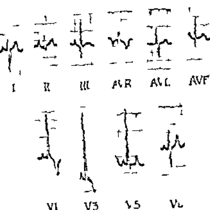


FIGURE VII–6 Origin of both great vessels from the right ventricle and pulmonary stenosis (same patient as in Figure VII–4) Infant

the catheter usually enters the aorta with ease If the pulmonary artery can be catheterized, the pulmonary pressure is low The oxygen content of the blood in the aorta and in the pulmonary artery is virtually the same

Angiocardiography shows that the aorta fills simultaneously with a small pulmonary artery It is of interest that the case which illustrates this malformation was misdiagnosed because it was thought that the aorta was not transposed The aorta did *not* appear to lie as far anteriorly as is usual when the aorta is transposed Nevertheless, careful examination of the aorta relative to the left ventricle in the lateral films taken after the left ventricle has filled suggests that the aorta lies anterior to the left ventricle (see Figure VII–7) Selective angiocardiography, with dye placed in the pulmonary artery, should clearly demonstrate the left auricle, the left ventricle, and the anterior position of the aorta

DIAGNOSIS

The diagnosis is based upon the finding of persistent cyanosis, which appears at an early age and is associated with severe dyspnea, absence of attacks of paroxysmal dyspnea the occurrence of episodes of deepening cyanosis, which often progress to loss of consciousness and a heart of characteristic contour The absence of fullness of the pulmonary conus and the concave curve of the cardiac silhouette at the base of the heart to the left of the sternum show that the pulmonary artery is diminutive, misplaced, or absent In the left anterior-oblique position both ventricles are seen to be greatly enlarged and the pulmonary window is wide and clear There is electrocardiographic evidence of extreme right ventricular hypertrophy



Left anterior oblique position



Right anterior oblique position

FIGURE VII–5 Origin of both great vessels from the right ventricle and pulmonary stenosis (same patient as in Figure VII–4) Infant

called right ventricular 'strain The P waves are often high and peaked (see Figure VII–6)

SPECIAL TESTS

The circulation time (arm to tongue) is short

The oxygen saturation of the arterial blood is low and falls still further with exercise

Cardiac catheterization reveals systemic pressure in the right ventricle and

DIFFERENTIAL DIAGNOSIS

This malformation requires differentiation from a defective development of the right ventricle with an intact ventricular septum and pulmonary stenosis, from a tetralogy of Fallot with severe pulmonary stenosis or functional pulmonary atresia, from truncus arteriosus with reduced pulmonary blood flow, and from a complete transposition of the great vessels with pulmonary stenosis

Defective development of the right ventricle with an intact ventricular septum is readily *confused* with the malformation under discussion When there is a defective development of the right ventricle but a normally placed aorta, the infant does not suffer from attacks of paroxysmal dyspnea If, however, an giocardiography is performed, although the pulmonary artery will be seen to arise from the right ventricle, the aorta will not be visualized until after the left auricle and the left ventricle have filled

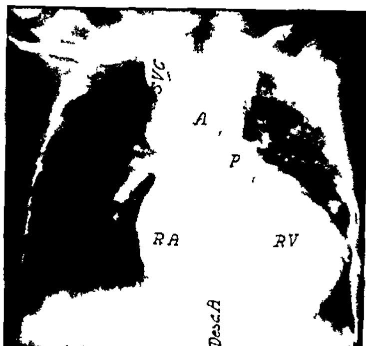
A tetralogy of Fallot with anatomical or functional pulmonary atresia causes the heart to remain small and in the left anterior-oblique position it appears to be globular In contrast to this, when both great vessels arise from the right ventricle, that chamber is always enlarged, if the duration of life permits, the left ventricle will also become enlarged because of the difficulty in the expulsion of blood from the left ventricle

Truncus arteriosus in which both ventricles are normally formed and the circulation to the lungs is through the enlarged bronchial arteries, is differentiated by x ray examination The *truncus* is usually larger than the aorta and the aortic knob lies at an abnormally high level The heart is also relatively larger and extends out as a shelf to the left of the sternum The infant usually does better than in the malformation under discussion

Complete transposition of the great vessels combined with pulmonary stenosis may in early infancy closely resemble the malformation under discussion When the great vessels are transposed, the vascular markings extend to the periphery of the lung fields

TREATMENT

Treatment is unsatisfactory A systemic pulmonary anastomosis increases the volume of venous blood directed to the lungs but there is usually such great difficulty in the expulsion of blood from the left ventricle that the infant dies from pulmonary edema The creation of an auricular defect *may lessen this* difficulty but after operation the strain on the circulation is so great that the child dies from cardiac failure



Simultaneous visualization of the aorta and the pulmonary artery



Superimposition of the aorta and pulmonary artery

FIGURE VII-7 Origin of both great vessels from the right ventricle and pulmonary stenosis (same patient as in Figure VII-4) Infant

also from a complete transposition of the great vessels with pulmonary stenosis

Treatment is unsatisfactory because any increase in the circulation to the lungs increases the volume of blood returned to the left ventricle Usually the difficulty in the expulsion of the blood from the left ventricle is so great that the infant dies of pulmonary edema Without operation the child usually dies of anoxemia or cardiac failure Therefore, either with or without operation the prognosis is poor Most patients die in early childhood

References

1 Mehrizi A (To be published)

2 Abbott M E Atlas of Congenital Cardiac Disease New York American Heart Association 1936

3 Lev M and O Saphir Transposition of the large vessels J Tech Methods 17 1 6–16 1937

4 Witham C A Double outlet right ventricle Am Heart J 53 928–939 1957

Relief of the pulmonary stenosis under direct vision may improve the circulation to the lungs, but, if the pulmonary artery is widely opened, the blood will be pumped to the lungs under systemic pressure Furthermore, because of the difficulty in the expulsion of blood from the left ventricle, the increased circulation to the lungs usually precipitates pulmonary edema

PROGNOSIS

Prognosis is poor Most patients die in infancy or early childhood Nevertheless, the author knows of three patients who lived to adult life and died at operation A systemic pulmonary anastomosis combined with the creation of an *auricular defect may lessen the incapacity but frequently leads to acute pulmonary* edema or intractable cardiac failure

SUMMARY

The origin of both great vessels from the right ventricle, with or without pulmonary stenosis, is a very serious malformation The fact that the aorta arises from the right ventricle means that venous blood is pumped directly into the systemic circulation In the presence of pulmonary stenosis little blood reaches the lungs for oxygenation In the absence of pulmonary stenosis blood is ejected to the lungs under systemic pressure, furthermore, owing to the difficulty in the expulsion of blood from the left ventricle, the pressure in the left auricle is increased This also leads to pulmonary hypertension Consequently pulmonary *hypertension is severe The condition soon leads to pulmonary edema*

The outstanding clinical findings are intense cyanosis, dyspnea, and absence of attacks of paroxysmal dyspnea The child squats when tired

The cardiac findings are similar to those of a tetralogy of Fallot with great over riding of the aorta The pulmonic second sound is accentuated but not reduplicated The x ray shows moderate *cardiac enlargement and a concave curve* at the base of the heart The lung fields are usually clear Viewed in the left anterior oblique position both ventricles are enlarged and the pulmonary window is wide and clear

The electrocardiogram shows a right axis deviation and evidence of extreme right ventricular hypertrophy

Angiocardiography shows that both vessels are filled from the right ventricle

The condition requires differentiation from defective development of the right ventricle with an intact ventricular septum and pulmonary stenosis a tetralogy of Fallot, a truncus arteriosus with reduced pulmonary blood flow, and

also from a complete transposition of the great vessels with pulmonary stenosis

Treatment is unsatisfactory because any increase in the circulation to the lungs increases the volume of blood returned to the left ventricle Usually the difficulty in the expulsion of the blood from the left ventricle is so great that the infant dies of pulmonary edema Without operation the child usually dies of anoxemia or cardiac failure Therefore, either with or without operation the prognosis is poor Most patients die in early childhood

References

1 Mehrizi A (To be published)

Abbott M E Atlas of Congenital Cardiac Disease New York American Heart Association 1936

3 Lev M and O Saphir Transposition of the large vessels J Tech Methods 17 1 6–162 1937

4 Witham C A Double outlet right ventricle Am Heart J 53 9 8–939 1957

CHAPTER VIII

TRICUSPID ATRESIA AND DEFECTIVE DEVELOPMENT OF THE RIGHT VENTRICLE

TRICUSPID ATRESIA *means that there is no tricuspid orifice, hence the* blood cannot reach the right ventricle in the normal manner, consequently *the right ventricle is always abnormally small* For this reason the two conditions are usually part and parcel of the same malformation The right ventricle may be absent or it may persist as a blind sac which does not communicate with the circulation Sometimes the primitive outflow tract develops as a small chamber which receives its blood from the left ventricle through a defect in the ventricular septum [1] The great vessels may or may not be transposed In all instances it is the diminutive size of the right ventricle which offers the clue to the nature of the underlying malformation The small size of the right ventricle can be seen by x ray and fluoroscopy and is confirmed by the electrocardiographic finding of a left ventricular preponderance in the precordial leads

Section A is concerned with tricuspid atresia combined with defective development of the right ventricle in which the great vessels are normally placed Section B is concerned with tricuspid atresia when there is a small right ventricle from which the aorta arises

A *Tricuspid Atresia Combined with Defective Development of the Right Ventricle and with Normally Placed Great Vessels*

NATURE OF THE MALFORMATION

The essential feature of this malformation is the atresia of the tricuspid valve, its orifice is represented by a dimple in the base of the right auricle In addition, there is defective development of the right ventricle The right ventricle may be absent, there may be a rudimentary chamber which has no demonstrable connection with the circulation, or in rare instances there may be extreme hypoplasia of the tricuspid valve and the right ventricle may exist as a diminutive chamber which is filled with a blood clot In each of these conditions the right ventricle fails to function as an integral part of the heart When the tricuspid valve is atretic or markedly hypoplastic and the right ventricle is a non functioning chamber, there is inevitably some malformation of the pulmonary orifice A

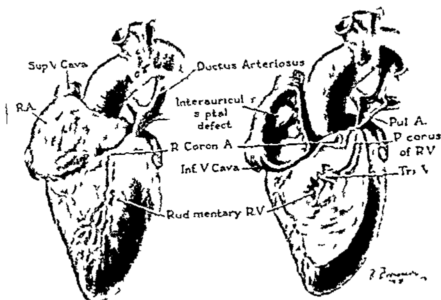


FIGURE VIII–1 Tricuspid atresia a non functioning right ventricle and a gross auricular septal defect (same patient as in Figure VIII–8) Infant

The diminutive size of the right ventricle caused marked distortion of the specimen on fixation

functioning pulmonary artery cannot be given off a non functioning chamber It follows that the pulmonary artery is either atretic or occupies an abnormal position Usually there is pulmonary atresia and the blood reaches the lungs through the ductus arteriosus, as shown in Figure VIII–1

In some instances the primitive bulbus cordis develops into an outflow chamber which receives its blood from the left ventricle through a defect in the ventricular septum Under such circumstances the abnormality concerns the tricuspid valve and the development of the right ventricle, the pulmonary artery is usually normally formed and of relatively normal size Furthermore, the circulation to the lungs is proportionately better when there is a small right ventricle than when the right ventricle is non functioning Such is the usual structure of the heart in patients with tricuspid atresia who survive for more than two years, the condition may be compatible with life for a number of years Figure VIII–2 illustrates this type of anomaly in a four year-old child, and Figure VIII–3 is a drawing of the heart of a patient who died at operation at thirty five years of age

The atresia of the tricuspid orifice and the non functioning right ventricle

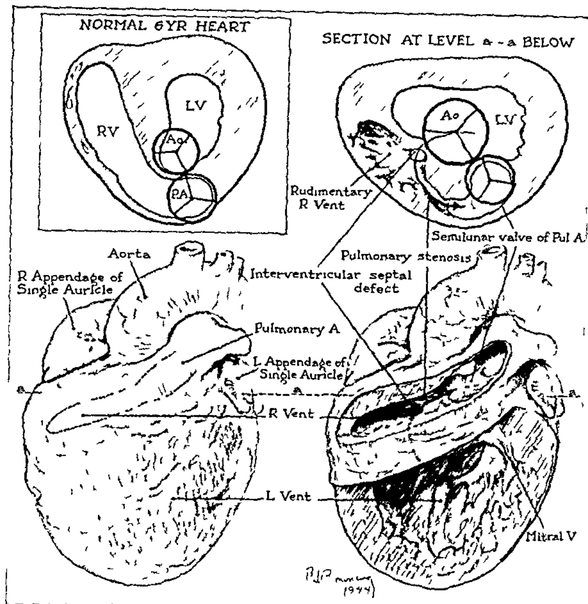


FIGURE VIII-2 Tricuspid atresia and a rudimentary right ventricle which receives blood from the left ventricle through a small ventricular septal defect (same patient as in Figure VIII-11) Child

mean that the blood which enters the right auricle through the superior and inferior venae cavae cannot leave that chamber in the normal manner The blood from the right auricle must escape by way of the left auricle This renders inevitable some defect in the auricular septum The foramen ovale may be normal in structure and covered by a valve which is not completely sealed, or there may be a gross defect in the auricular septum In either case there are two auricles and one ventricle, which constitutes a triloculate heart In some cases the auricular septum is entirely lacking except for a few strands of tissue Under

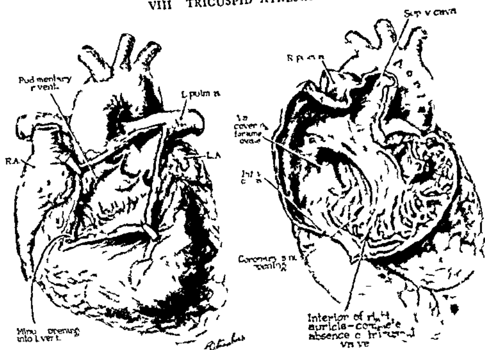


Figure VIII–3 Tricuspid atresia with the pulmonary artery arising from a rudimentary right ventricle Adult

such circumstances the two auricles function as a single chamber There is but one auricle and one ventricle, functionally there is a biloculate heart In order to make a clinical diagnosis it is necessary first to establish the diagnosis of tricuspid atresia and a diminutive or absent right ventricle and then to analyze the condition within the auricles

COURSE OF THE CIRCULATION

During fetal life the foramen ovale and the ductus arteriosus are normally patent Inasmuch as the tricuspid orifice is atretic and the right ventricle does not function all the blood from the right auricle must flow through the foramen ovale into the left auricle and thence to the left ventricle The left ventricle pumps the blood through the aorta to the body of the fetus and through the ductus arteriosus into the branches of the pulmonary artery and thence to the lungs Inasmuch as the lungs do not function during fetal life, the normal flow of blood to the lungs is minimal, hence the absence of the right ventricle places little strain upon the fetal circulation The heart at birth is of normal size Figure VIII–4 shows the course of the fetal circulation when there is tricuspid atresia and pulmonary atresia Figure VIII–5 shows the course of the fetal circulation

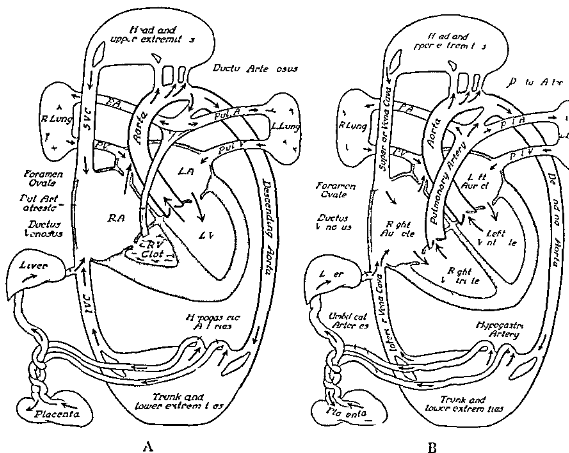


FIGURE VIII–4 Fetal circulation (A) Non functioning right ventricle and a well formed auricular septum and (B) normal heart

when in spite of the tricuspid atresia the pulmonary artery arises from the in fundibular chamber of the right ventricle

After birth some circulation to the lungs must be established, usually this is by way of the ductus arteriosus It is, of course, possible to have other pathways, such as a transposed pulmonary artery or a truncus arteriosus Such conditions, however, represent additional anomalies (discussed in Section B and in Chapter XIV, respectively), whereas the ductus arteriosus represents the persistence of a normal fetal pathway

As long as the ductus arteriosus remains patent, the blood from the aorta passes through it to the pulmonary artery The pulmonary artery proximal to the ductus arteriosus usually persists as a small non functioning vessel The main branches of the pulmonary artery beyond the ductus arteriosus are normally developed The course of the circulation is shown in Diagram VIII–1

All the blood which enters the right auricle flows into the left auricle and thence to the left ventricle Therefore, the blood in the left ventricle is a mix

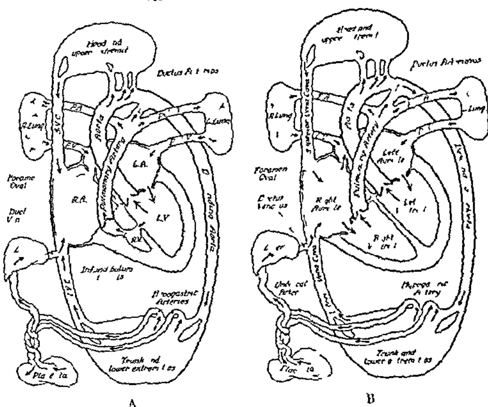


FIGURE VIII-5 Fetal circulation. (A) Tricuspid *atresia and stenosis* of the infundibulum and (B) *normal heart*

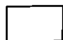
ture of oxygenated and venous blood The blood in the left ventricle is pumped out through the aorta to the systemic circulation, blood from the aorta also flows through the ductus arteriosus to the pulmonary artery and thence to the lungs The oxygenated blood from the lungs is returned by the pulmonary veins to the left auricle The blood from the systemic circulation is returned by the superior vena cava and the inferior vena cava to the right auricle, thence it flows to the left auricle There the cycle starts again

Absence of the auricular septum does not greatly alter the course of the circulation The blood from the superior vena cava and the inferior vena cava enters the right side of the common auricle, and that from the pulmonary veins enters the left side All the blood from both auricles flows through the mitral valve to the left ventricle and out by way of the aorta to the systemic circulation and through the ductus arteriosus to the pulmonary arteries and the lungs, as shown in Diagram VIII-2

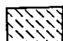
When the right ventricle remains as a rudimentary chamber which receives

Diagram VIII–1

Head and upper extremities
Ductus Arteriosus
R. Lung
S.V.C.
P.A.
Aorta
Pul. A.
L. Lung
PV
Pul V
L.A.
R.A.
L.V.
R.V.
Remnant of Pul Art
Descending Aorta
I.V.C.
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis

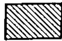

Venous blood

Diagram VIII–1

Marked hypoplasia of the tricuspid orifice and a non functioning right ventricle combined with a well formed auricular septum pulmonary atresia and a patent ductus arteriosus

In this malformation there are two auricles and a single ventricle the left The right ventricle, even if present is diminutive in size and does not function The tricuspid valve is atretic or markedly hypoplastic In addition, there is pulmonary atresia hence the only way for the blood to reach the lungs is by way of the ductus arteriosus The auricular septum is well formed but there is either a small defect or a functionally patent foramen ovale

The abnormality of the tricuspid valve and the diminutive right ventricle prevent the normal flow of blood from the right auricle to the right ventricle Whatever blood does enter the right ventricle cannot escape and becomes clotted The only way for the blood to circulate is from the right auricle, through the foramen ovale, into the left auricle Thence it flows into the left ventricle and is pumped out by way of the aorta to the systemic circulation and through the ductus arteriosus to the lungs The blood from the systemic circulation is returned in the normal fashion by the superior vena cava and the inferior vena cava to the right auricle and thence to the left auricle Inasmuch as there is pulmonary atresia the pressure in the pulmonary circulation is low and blood will flow from the aorta through the ductus arteriosus to the lungs This is the only way by which the blood can reach the lungs The blood from the lungs is returned in the normal fashion by the pulmonary veins to the left auricle

The volume of blood shunted through the ductus arteriosus is small Consequently only a small amount of blood reaches the lungs for oxygenation and a correspondingly small amount of oxygenated blood is returned to the left auricle Hence the pressure in the left auricle remains low This aids the flow of blood from the right auricle to the left auricle The small amount of oxygenated blood returned to the left auricle and the large volume of venous blood shunted from the right auricle to the left auricle produce cyanosis

As the infant grows and the ductus arteriosus undergoes normal obliteration the supply of blood to the lung decreases Cyanosis becomes progressively more intense With the closure of the ductus arteriosus the condition usually becomes incompatible with life Death commonly occurs between the ages of three and five months

Clinical diagnosis is made by x ray or fluoroscopic examination The shape of the heart is characteristic The absence of the right ventricle causes the left ventricle to enlarge In the anterior posterior view there is absence of the shadow normally cast by the pulmonary conus In the left anterior-oblique position the enlargement of the heart is seen to be due to the left ventricle the right ventricle does not project forward beyond the margin of the aorta The pulmonary window is unusually clear Murmurs if present, are systolic in time and are of no diagnostic aid The electrocardiogram shows a left axis deviation and evidence of left ventricular dominance

DIAGRAM VIII–2

Head and upper extremities

Ductus Arteriosus

R Lung

P.A.

S.V.C.

Aorta

Pul. A

L Lung

PV

Pul V

L.A.

R.A.

L.V.

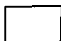
Remnant of Pul Art

R.V.

Descending Aorta

I.V.C.

Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis

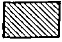

Venous blood

Diagram VIII-2

Tricuspid atresia and a non functioning right ventricle combined with a gross auricular septal defect pulmonary atresia and a patent ductus arteriosus

In this malformation in addition to the non functioning right ventricle and the pulmonary atresia there is such a gross defect in the auricular septum that the two auricles function as a single chamber Each auricle occupies its normal position There is either tricuspid atresia or marked hypoplasia of the tricuspid orifice

The blood from the right auricle cannot leave by its normal pathway it can escape only by way of the mitral orifice into the left ventricle The blood from the left ventricle is pumped out through the aorta to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle Thence it again flows into the left auricle and the left ventricle Inasmuch as there is pulmonary atresia, the pressure in the pulmonary circulation is low So long as the ductus arteriosus is patent some blood will flow from the aorta through the ductus arteriosus to the pulmonary artery *This is* the only means by which the blood can reach the lungs The blood from the lungs is returned by the pulmonary veins in the normal fashion to the left auricle

Thus the left auricle receives oxygenated blood from the lungs and venous blood from the systemic circulation this mixture of arterial and venous blood flows into the left ventricle and is pumped into the aorta The greater portion of the blood in the aorta is pumped into the systemic circulation only a relatively small volume of blood reaches the lungs for oxygenation It follows that only a small volume of oxygenated blood is returned to the left auricle where it is mixed with a large volume of venous blood received from the right auricle Cyanosis is intense As the ductus arteriosus closes the condition becomes incompatible with life

Clinical diagnosis is made by x ray or fluoroscopic examination and confirmed by the electrocardiographic findings Inasmuch as the duration of life is short, the heart is comparatively small The shape of the heart is characteristic The right ventricle is absent the left ventricle which does the work of both ventricles is enlarged In the anterior posterior view there is an absence of the fullness of the pulmonary conus In the left anterior-oblique position there is an absence of enlargement of the right ventricle The gross defect in the auricular septum increases the pressure in the right auricle and causes dilatation of the superior vena cava A systolic murmur may or may not be present such murmurs are of no diagnostic aid During the neonatal period the electrocardiogram may show a balanced axis subsequently the electrocardiogram will show a left axis deviation but the unipolar precordial leads show evidence of left ventricular dominance which is present at birth and conspicuous in V_4 as well as in V_5 and V_6

its blood from the left ventricle, the circulation is altered only by the volume c blood which is pumped from the left ventricle into the rudimentary right ven tricle and thence through the pulmonary artery to the lungs In such cases, al though the pulmonary blood flow is reduced, life is not dependent upon th patency of the ductus arteriosus Consequently this condition may be compatibl with life for a number of years The course of the circulation is shown in Dia gram VIII–3

PHYSIOLOGY OF THE MALFORMATION

The altered structure of the heart causes difficulty in the expulsion of bloo from the right auricle and in the direction of blood to the lungs Consequentl there is always a reduction in the pulmonary blood flow except when the mal formation occurs in combination with a transposition of the great vessels (se Section B) The reduction in the pulmonary blood flow, combined with the ad mixture of venous and arterial blood in the left ventricle, causes a marked re duction in the oxygen saturation of the arterial blood The pulmonary stenosi reduces the pressure in the pulmonary artery, hence the malformation does no injure the lungs When there is pulmonary atresia, life is dependent on th patency of the ductus arteriosus or on the development of collateral circulation

CLINICAL FINDINGS

Cyanosis is persistent and usually dates from birth In this malformation only a small amount of blood reaches the lungs for oxygenation and therefore only a small amount of oxygenated blood is returned to the left auricle, where it is mixed with a large volume of venous blood which has been returned from the systemic circulation to the right auricle and thence to the left auricle This mix ture of venous and arterial blood passes from the left auricle to the left ventricle and is pumped out into the aorta Therefore cyanosis is intense and of uniform distribution

Clubbing of the fingers and toes develops at an early age

Difficulty in feeding is a common complaint Most infants with this malfor mation gain weight very slowly *Failure to gain* is an ominous sign

The occurrence of attacks of paroxysmal dyspnea is usually extremely seri ous, such attacks occur as the ductus arteriosus undergoes obliteration More over, life is usually dependent upon the patency of the ductus arteriosus In this malformation there is real danger that the first attack of paroxysmal dyspnea will be the last

If the infant is able to establish sufficient circulation to the lungs, he outgrows the attacks of paroxysmal dyspnea between eighteen months and two years of age, thereafter he starts to improve

Squatting is quite as common a habit for these children as for those with a tetralogy of Fallot Indeed, their exercise tolerance varies in a similar manner to that of patients with a tetralogy of Fallot and their general behavior is also closely similar

The blood pressure is narrow and may be difficult to obtain The pulses in the arm and the leg are of equal strength

The liver may or may not be enlarged Pulsations at the margin are frequently palpable Even when the liver is normal in size, the heart beat may be readily counted by placing one s hand against the margin of the liver in the anterior axillary line When such pulsations are palpable, the opening between the two auricles is small

CARDIAC FINDINGS

The heart is usually slightly enlarged Generally the apex thrust is palpable beyond the mid clavicular line

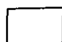
The quality of the second sound is remarkably pure Since there is only *one great vessel*, there can be no reduplication of the second sound It is to be emphasized that it is the *purity* of this sound, not the location, which is of significance The pulmonic second sound may be louder than the aortic second sound This does not mean that the sound is produced by the closure of the pulmonary valves It means only that the closure of the aortic valve is better heard to the left of the sternum than to the right

Murmurs may or may not be present The only abnormal openings which could cause murmurs are the ductus arteriosus and a defect in the auricular septum The defect between the auricles may be so large that the two auricles function as a single chamber The *ductus arteriosus* is not merely a connection between the two circulations but is the main pathway to the lesser circulation Inasmuch as the occurrence of a murmur depends upon the size of the openings and the relative pressure in the two circulations, there may or may not be a murmur For this reason murmurs are of no diagnostic importance

The structure of the auricular septum is adduced from the size of the liver and the presence or absence of pulsations at the margin of the liver

A well formed auricular septum when combined with a non functioning right ventricle, causes the heart to function as a triloculate one, that is, two auri

Diagram VIII–3



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

Diagram VIII-3

Tricuspid atresia and a gross auricular septal defect combined with defective development of the right ventricle infundibular stenosis and a small ventricular septal defect

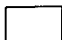
The essential features of this malformation are tricuspid atresia and defective development of the right ventricle Inasmuch as the tricuspid valve is atretic the blood must flow from the right auricle to the left auricle it follows that either the foramen ovale is held open by the high pressure in the right auricle or there is a defect in the auricular septum The only way for the blood to enter the right ventricle is from the left ventricle through a ventricular septal defect There may also be infundibular stenosis of the right ventricle which places an additional obstruction to the flow of blood through the right ventricle to the pulmonary artery

The blood flows from the right auricle through the defect in the auricular septum into the left auricle, where it meets the blood which is returned by the pulmonary veins to the left auricle This mixture of venous blood from the right auricle and oxygenated blood from the left auricle flows through the mitral valve into the left ventricle From the left ventricle most of the blood is pumped out through the aorta to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle A small volume of blood is forced through the ventricular septal defect into the diminutive right ventricle from which it is pumped into the pulmonary artery This is the only blood which reaches the lungs The blood which reaches the lungs is returned in the normal fashion to the left auricle There the cycle starts again

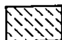
Clinical diagnosis is based on the contour of the heart In the anterior posterior position the infundibular stenosis causes an absence of fullness of the pulmonary conus The small size of the right ventricle is seen in the left anterior-oblique position Owing to the complete admixture of the oxygenated and venous blood in the left auricle and the small volume of blood which reaches the lungs for oxygenation cyanosis is intense After the closure of the ductus arteriosus the blood can still reach the lungs by way of the pulmonary artery therefore, the condition is compatible with life for a longer time than when tricuspid atresia is combined with a non functioning right ventricle The electrocardiogram shows a left axis deviation and evidence of left ventricular preponderance

Diagram VIII–3

Head and upper extremities
R Lung
L Lung
SVC
PA
PV
Aorta
Pul. A
Pul V
Pulmonary Artery
LA
RA
LV
RV
Descending Aorta
IVC
Infundibulum Stenosis
Defect in Interventricular Septum
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis

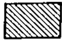

Venous blood

Diagram VIII–3

Tricuspid atresia and a gross auricular septal defect combined with defective development of the right ventricle infundibular stenosis and a small ventricular septal defect

The essential features of this malformation are tricuspid atresia and defective development of the right ventricle Inasmuch as the tricuspid valve is atretic, the blood must flow from the right auricle to the left auricle it follows that either the foramen ovale is held open by the high pressure in the right auricle or there is a defect in the auricular septum The only way for the blood to enter the right ventricle is from the left ventricle through a ventricular septal defect There may also be infundibular stenosis of the right ventricle which places an additional obstruction to the flow of blood through the right ventricle to the pulmonary artery

The blood flows from the right auricle through the defect in the auricular septum into the left auricle where it meets the blood which is returned by the pulmonary veins to the left auricle This mixture of venous blood from the right auricle and oxygenated blood from the left auricle flows through the mitral valve into the left ventricle. From the left ventricle most of the blood is pumped out through the aorta to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right auricle A small volume of blood is forced through the ventricular septal defect into the diminutive right ventricle from which it is pumped into the pulmonary artery This is the only blood which reaches the lungs The blood which reaches the lungs is returned in the normal fashion to the left auricle There the cycle starts again

Clinical diagnosis is based on the contour of the heart In the anterior posterior position the infundibular stenosis causes an absence of fullness of the pulmonary conus The small size of the right ventricle is seen in the left anterior-oblique position Owing to the complete admixture of the oxygenated and venous blood in the left auricle and the small volume of blood which reaches the lungs for oxygenation cyanosis is intense After the closure of the ductus arteriosus the blood can still reach the lungs by way of the pulmonary artery therefore, the condition is compatible with life for a longer time than when tricuspid atresia is combined with a non functioning right ventricle The electrocardiogram shows a left axis deviation and evidence of left ventricular preponderance

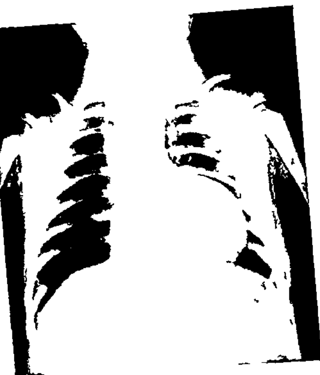
cles and a single ventricle If the opening in the auricular septum is relatively small, that is smaller than that of the normal tricuspid orifice, it is difficult for the right auricle to expel the blood The condition is functionally similar to that of an acquired tricuspid atresia in which it is difficult for the auricle to expel the blood through the stenosed tricuspid valve Both conditions are characterized by the same clinical phenomenon namely, the auricular pulsations are transmitted to the liver and cause presystolic pulsations at its margin

The differentiation of a presystolic pulsation from a systolic pulsation may be difficult, if not impossible, to determine, owing to the rapid rate of the infant s heart The two conditions can, however, be distinguished by the size of the right ventricle and not infrequently by the size of the liver The size of the right ventricle is determined by fluoroscopy and that of the liver by palpation A systolic pulsation occurs in association with a great dilatation of the right ventricle, tricuspid insufficiency, and engorgement of the liver In contrast to this, a presystolic pulsation in infancy is associated with a tricuspid atresia or marked hypoplasia, which in turn occurs in conjunction with a diminutive or an absent right ventricle and a liver of normal size It follows that in this malformation a pulsating liver of normal size combined with the absence of right ventricular enlargement is presumptive evidence of tricuspid atresia and a well formed auricular septum with a relatively small defect

A gross defect in the auricular septum, when sufficiently large to permit the free flow of blood from the right auricle to the left auricle, relieves the pressure in the right auricle, hence the liver does not pulsate

X RAY AND FLUOROSCOPIC FINDINGS

The absence of the right ventricle causes a characteristic change in the contour of the heart In the anterior posterior position the absence of the right ventricle is suggested by the absence of the shadow cast by the pulmonary conus of the right ventricle The upper margin of the cardiac shadow to the left of the sternum, instead of showing its usual convexity, is concave (Figures VIII–6, 7, 8) In the left anterior-oblique position the diminutive size of the right ventricle is indicated by the fact that the cardiac shadow does not project toward the anterior chest wall beyond the shadow cast by the aorta (see Figures VIII–6 and 7) The left ventricle, on the other hand, is enlarged and a considerable degree of rotation is required for the left ventricle to clear the spinal column Therefore, in spite of the absence of the right ventricle, the heart appears to be slightly enlarged, the enlargement, however, is entirely due to the increased size of the left ventricle



Anterior posterior position



Left anterior-oblique position

Figure viii-6 Tricuspid atresia, a non functioning right ventricle, and a well formed auricular septum Infant

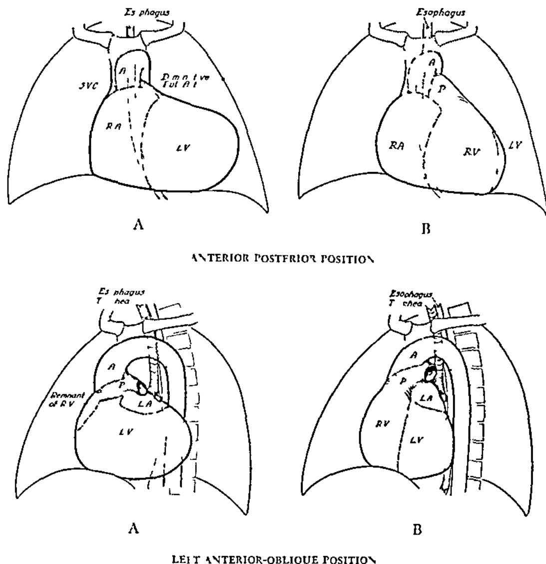


FIGURE VIII–7 (A) Tricuspid atresia and a non functioning right ventricle and (B) normal heart Infant

The diminutive size of the pulmonary artery causes the shadow cast by the great vessels in the anterior posterior position to be narrow Inasmuch as there is but a single vessel, upon rotation of the patient into the left anterior oblique position there is no increase in the width of this shadow In addition, the diminutive pulmonary artery renders the pulmonary window abnormally clear

Absence of the auricular septum results in a single auricle Usually the portion of the auricle which structurally represents the right auricle is more distensible and dilates more than the left side In early infancy the increased pressure on the right side of the auricle causes dilatation of the superior vena cava,

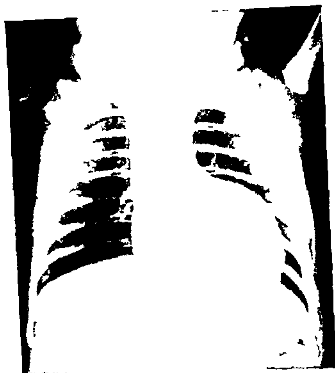


FIGURE VIII-8 Tricuspid atresia a non functioning right ventricle, and a gross auricular septal defect (same patient as in Figure VIII-1) Infant

which is readily seen by x ray and fluoroscopy, as illustrated in Figures VIII-9 and 10

This malformation may be compatible with life for a number of years When this occurs, it is common to find that the pulmonary artery arises from the outflow tract of the right ventricle in the normal manner and that the rudimentary right ventricle receives its blood from the left ventricle through a defect in the ventricular septum

If the infant survives to childhood, as the patient grows the diaphragm descends and the contour of the heart comes to resemble that of a tetralogy of Fallot Generally, however, the cardiac contour is more nearly square than in the tetralogy of Fallot This is due to the straight margin of the right auricle and to the slight fullness in the region of the pulmonary conus caused by the rudimentary outflow tract of the right ventricle and the relatively normal size of the pulmonary artery (see Figures VIII-11 and 12)

The hilar shadows may become accentuated as collateral circulation develops early and increases rapidly to compensate for the reduced flow through the main

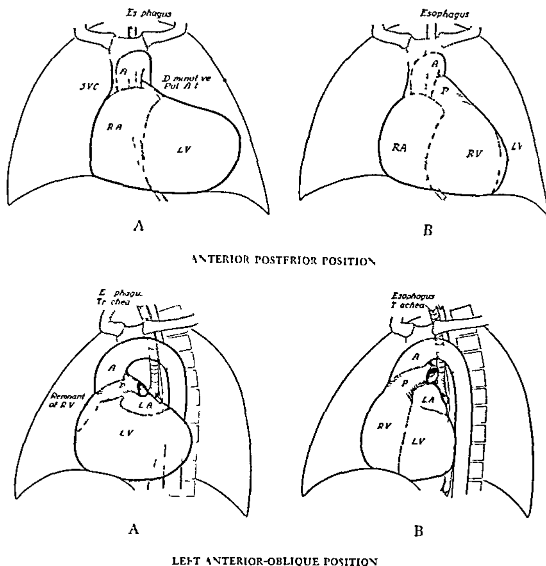
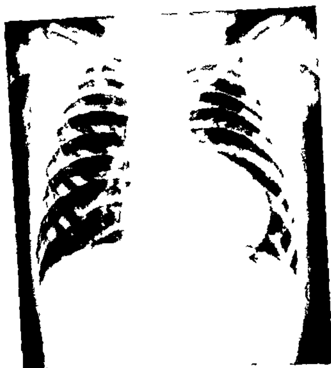


FIGURE VIII–7 (A) Tricuspid atresia and a non functioning right ventricle and (B) normal heart Infant

The diminutive size of the pulmonary artery causes the shadow cast by the great vessels in the anterior posterior position to be narrow Inasmuch as there is but a single vessel, upon rotation of the patient into the left anterior oblique position there is no increase in the width of this shadow In addition, the diminutive pulmonary artery renders the pulmonary window abnormally clear

Absence of the auricular septum results in a single auricle Usually the portion of the auricle which structurally represents the right auricle is more distensible and dilates more than the left side In early infancy the increased pressure on the right side of the auricle causes dilatation of the superior vena cava,



Anterior posterior position



Left anterior oblique position

FIGURE VIII-11 Tricuspid atresia and a *rudimentary right* ventricle (same patient as in Figure VIII-2) Child

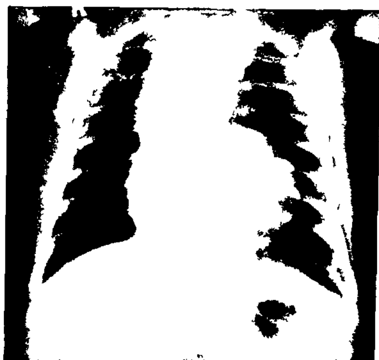


Figure viii–9 Tricuspid atresia, a non functioning right ventricle, and a single auricle Infant

Note the dilatation of the superior vena cava

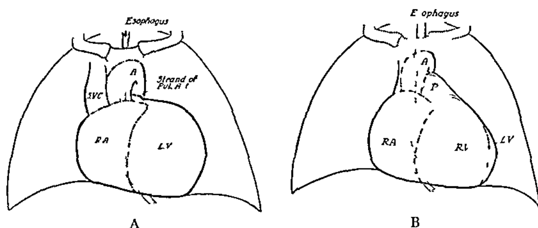
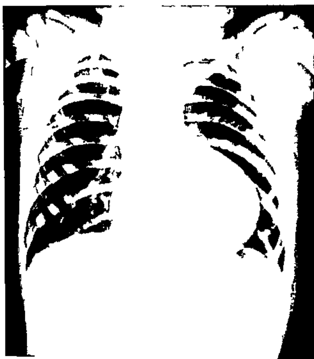


Figure viii–10 (A) Tricuspid atresia, a non functioning right ventricle, a single auricle, and a dilated superior vena cava and (B) normal heart Infant

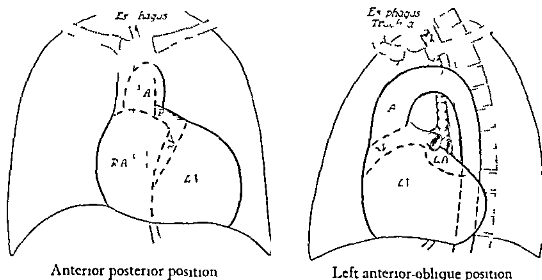


Anterior posterior position



Left anterior-oblique position

FIGURE VIII-11 Tricuspid atresia and a rudimentary right ventricle (same patient as in Figure VIII-2) Child



Anterior posterior position

Left anterior-oblique position

FIGURE VIII-12 Tricuspid atresia Child

pulmonary artery These shadows are an aggregate of small shadows and do not pulsate The periphery of the lungs remains clear

In the left anterior oblique position, as the heart drops down, the anterior border of the heart and the aorta form an almost vertical line, which is frequently cut in toward the spinal column at the level of the diaphragm The aortic shadow is narrow and the pulmonary window is clear (see Figures VIII–11 and 12)

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiographic findings are of diagnostic importance The finding of a left axis deviation in the standard leads, combined with evidence of left ventricular hypertrophy in the unipolar precordial leads, is always suggestive of tricuspid atresia Occasionally in young infants the standard leads may show a balanced axis, or in rare instances there may be even a right axis deviation The unipolar precordial leads, however, will uniformly show evidence of dominance of the left ventricle both in V_1 and in V_5 and V_6 (see Figure VIII–13) this finding is present at birth [2]

When there is but a small defect in the auricular septum, the P waves may be excessively tall and peaked, they are frequently over 5 mm and occasionally 10 mm in height These are the so called Himalayan P waves When pulsations are palpable at the margin of the liver in a patient who has Himalayan P waves in the electrocardiogram, the defect in the auricle is certain to be small

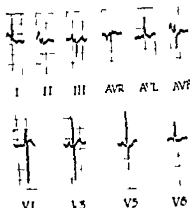


FIGURE VIII-13 Tricuspid atresia

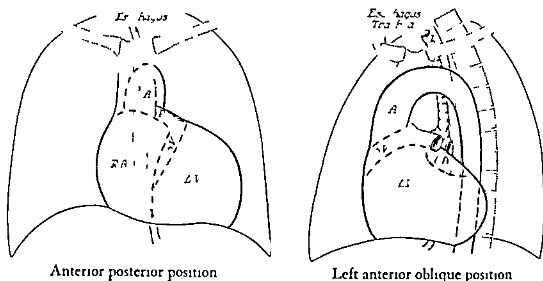
SPECIAL TESTS

The hematology is similar to that of other patients with persistent oxygen unsaturation of the arterial blood The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are all abnormally high Over the years these patients also show a reduction in the number of blood platelets and in the blood fibrinogen

The circulation time is usually shorter than normal as the blood passes directly from the right auricle to the left auricle and thence to the left ventricle and the systemic circulation

The oxygen saturation of the arterial blood is greatly reduced and falls still further with exercise The arterial oxygen saturation is lower when there is pulmonary atresia than when the pulmonary artery comes off the outflow tract of the right ventricle Under the former circumstance closure of the ductus arteriosus is generally fatal The determination of the oxygen saturation of the arterial blood is frequently the best guide in the evaluation of the severity of the condition Although the author has known one patient who lived to thirty five years of age in whom the resting oxygen saturation of the arterial blood was only 35 per cent such a saturation when obtained in an infant at rest is ominously low

Cardiac catheterization generally gives little information concerning the presence of tricuspid atresia, as it is frequently impossible to pass the catheter beyond the right auricle If, however, the diagnosis is wrong the catheter will pass through the tricuspid valve into the right ventricle, thereby demonstrating that the tricuspid valve is not atretic



Anterior posterior position

Left anterior oblique position

FIGURE VIII-12 Tricuspid atresia Child

pulmonary artery These shadows are an aggregate of small shadows and do not pulsate The periphery of the lungs remains clear

In the left anterior oblique position, as the heart drops down, the anterior border of the heart and the aorta form an almost vertical line, which is frequently cut in toward the spinal column at the level of the diaphragm The aortic shadow is narrow and the pulmonary window is clear (see Figures VIII-11 and 12)

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiographic findings are of diagnostic importance The finding of a left axis deviation in the standard leads, combined with evidence of left ventricular hypertrophy in the unipolar precordial leads, is always suggestive of tricuspid atresia Occasionally in young infants the standard leads may show a balanced axis, or in rare instances there may be even a right axis deviation The unipolar precordial leads, however, will uniformly show evidence of dominance of the left ventricle both in V_1 and in V_5 and V_6 (see Figure VIII-13) this finding is present at birth [2]

When there is but a small defect in the auricular septum, the P waves may be excessively tall and peaked, they are frequently over 5 mm and occasionally 10 mm in height These are the so-called 'Himalayan P waves When pulsations are palpable at the margin of the liver in a patient who has Himalayan P waves in the electrocardiogram, the defect in the auricle is certain to be small

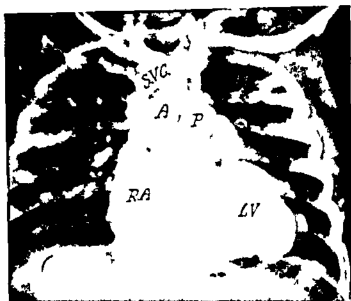




Figure VIII [illegible] Auricular septal defect with a rudimentary right ventricle and pulmonary stenosis Child

Anterior posterior and lateral films taken simultaneously

Angiocardiography is not of great diagnostic aid The left auricle is, of course, visualized immediately after the right auricle This finding does not, however, offer reliable evidence concerning the size of the auricular defect, it does not even aid in the differentiation of a gross defect in the auricular septum from patency of the valve covering the foramen ovale Immediately after the left auricle is filled, the dye enters the left ventricle and thereafter both the pulmonary artery and the aorta are filled Figure VIII–14 shows the dye in both the auricles Figure VIII–15, which was taken a fraction of a second later, shows dye in the left ventricle and also in both the pulmonary artery and the aorta The lateral view of Figure VIII–15 shows that the pulmonary artery lies anterior to the aorta

DIAGNOSIS

The diagnosis is based primarily on the finding of persistent cyanosis and electrocardiographic evidence of left ventricular hypertrophy This combination of findings always suggests a serious abnormality of the tricuspid valve and of the right ventricle Murmurs are of no diagnostic significance In infants, and occasionally in children, the contour of the heart may also be sufficiently char

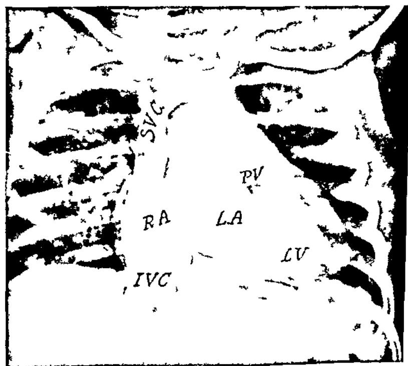


FIGURE VIII–14 Auricular septal defect with a rudimentary right ventricle and pulmonary stenosis Child

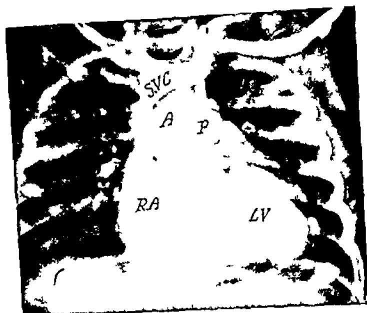




FIGURE VIII-15 Auricular septal defect with a rudimentary right ventricle and pulmonary stenosis Child

Anterior posterior and lateral films taken simultaneously

Angiocardiography is not of great diagnostic aid The left auricle is, of course, visualized immediately after the right auricle This finding does not, however, offer reliable evidence concerning the size of the auricular defect, it does not even aid in the differentiation of a gross defect in the auricular septum from patency of the valve covering the foramen ovale Immediately after the left auricle is filled, the dye enters the left ventricle and thereafter both the pulmonary artery and the aorta are filled Figure VIII–14 shows the dye in both the auricles Figure VIII–15, which was taken a fraction of a second later, shows dye in the left ventricle and also in both the pulmonary artery and the aorta The lateral view of Figure VIII–15 shows that the pulmonary artery lies anterior to the aorta

DIAGNOSIS

The diagnosis is based primarily on the finding of persistent cyanosis and electrocardiographic evidence of left ventricular hypertrophy This combination of findings always suggests a serious abnormality of the tricuspid valve and of the right ventricle Murmurs are of no diagnostic significance In infants, and occasionally in children, the contour of the heart may also be sufficiently char

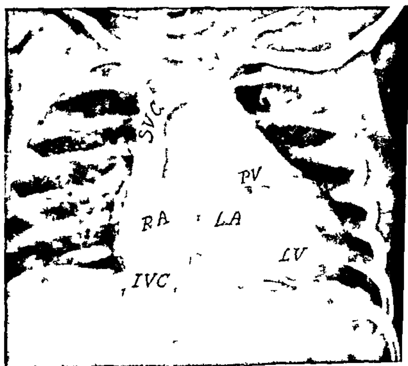


FIGURE VIII–14 Auricular septal defect with a rudimentary right ventricle and pulmonary stenosis Child

acteristic to suggest the diagnosis The significant x ray finding is the absence of the pulmonary conus in the anterior posterior position, combined with the absence of right ventricular enlargement in the left anterior-oblique position

DIFFERENTIAL DIAGNOSIS

In young infants the condition requires differentiation from a tetralogy of Fallot and from a single ventricle with pulmonary stenosis, and occasionally from a truncus arteriosus with markedly reduced pulmonary blood flow, and also from other malformations associated with defective development of the right ventricle, notably defective development of the right ventricle and pulmonary stenosis combined with an intact ventricular septum (see Chapter IX) and occasionally from tricuspid atresia combined with complete transposition of the great vessels with or without a single ventricle (see Section B)

A tetralogy of Fallot, especially in children, may closely resemble tricuspid atresia The clinical syndrome may be indistinguishable The two conditions are to be differentiated by the electrocardiographic findings In a tetralogy of Fallot there is usually a right axis deviation and evidence of right ventricular hypertrophy, whereas in a tricuspid atresia the electrocardiogram generally shows a left axis deviation and, regardless of age, almost invariably shows evidence of left ventricular dominance in V_1

A single ventricle with a rudimentary outlet chamber may require differentiation from a tricuspid atresia combined with defective development of the right ventricle In infancy the contours of the heart are similar in the two malformations when viewed in the left anterior-oblique position, but in the anterior posterior position their shapes are usually quite different In infancy a single ventricle with a rudimentary outlet chamber usually shows fullness of the pulmonary conus

As the infant grows to childhood and the diaphragm descends, the heart comes to occupy a more vertical position and the fullness caused by the rudimentary outflow chamber disappears The differential diagnosis is especially difficult when the electrocardiogram shows a left axis deviation, as in the case reported by Neill and Brink [2] The unipolar precordial leads are usually of aid in the differentiation of the two conditions In tricuspid atresia there is evidence from birth of left ventricular dominance in V_1, whereas in a single ventricle, although there may be a prominent R in V_5 or V_6, the left ventricular pattern is seldom pronounced in V_1

A single ventricle may occur in combination with pulmonary stenosis In

deed, the difference between a single ventricle and a rudimentary right ventricle which communicates with the left ventricle is primarily a matter of the formation of the ventricular septum In the former the outflow tract of the right ventricle appears as a pouch of the common ventricle, in the latter the septal wall is well formed and there is a relatively small defect through which the outflow tract communicates with the left ventricle (compare Figure VIII-2 with Figure XV-1) Cardiac catheterization will show that the common ventricle is entered directly from the right auricle

Truncus arteriosus with markedly reduced pulmonary blood flow may have a similar contour to the heart in the anterior posterior position, except that the aortic knob is usually more conspicuous In the left anterior-oblique position a truncus arteriosus shows enlargement of the right ventricle, this finding clearly distinguishes it from a non functioning right ventricle Further, the electrocardiogram shows evidence of hypertrophy of both ventricles and not a preponderant hypertrophy of the left ventricle Upon cardiac catheterization it will be possible to pass the catheter from the right auricle into the right ventricle and out into the aorta

Complete transposition of the great vessels may occur in combination with tricuspid atresia and defective development of the right ventricle (see Section B)

Defective development of the right ventricle and pulmonary stenosis with a normal tricuspid orifice and no over riding of the aorta is a rare malformation, nevertheless, it does occur (see Chapter IX) Occasionally this malformation is associated with a left axis deviation and evidence of left ventricular hypertrophy Usually, however, some evidence of right ventricular hypertrophy can be detected in V_1 or V_{3R} These findings give the clue to the diagnosis Cardiac catheterization will usually show high pressure in the right ventricle Angiocardiography will show that a small pulmonary artery arises from the right ventricle, but the aorta will not be simultaneously delineated

TREATMENT

In patients with tricuspid atresia combined with pulmonary stenosis or atresia the primary difficulty is lack of adequate circulation to the lungs Consequently these patients may be greatly helped by a Blalock Taussig operation or by a Potts anastomosis

There is, however, always the danger that the increased volume of blood returned from the lungs will raise the pressure in the left auricle to such an extent that the right auricle will have difficulty in expelling its blood to the left auricle

acteristic to suggest the diagnosis The significant x ray finding is the absence of the pulmonary conus in the anterior posterior position, combined with the absence of right ventricular enlargement in the left anterior oblique position

DIFFERENTIAL DIAGNOSIS

In young infants the condition requires differentiation from a tetralogy of Fallot and from a single ventricle with pulmonary stenosis, and occasionally from a truncus arteriosus with markedly reduced pulmonary blood flow, and also from other malformations associated with defective development of the right ventricle, notably defective development of the right ventricle and pulmonary stenosis combined with an intact ventricular septum (see Chapter IX) and occasionally from tricuspid atresia combined with complete transposition of the great vessels with or without a single ventricle (see Section B)

A tetralogy of Fallot, especially in children, may closely resemble tricuspid atresia The clinical syndrome may be indistinguishable The two conditions are to be differentiated by the electrocardiographic findings In a tetralogy of Fallot there is usually a right axis deviation and evidence of right ventricular hypertrophy, whereas in a tricuspid atresia the electrocardiogram generally shows a left axis deviation and, regardless of age, almost invariably shows evidence of left ventricular dominance in V_1

A single ventricle with a rudimentary outlet chamber may require differentiation from a tricuspid atresia combined with defective development of the right ventricle In infancy the contours of the heart are similar in the two malformations when viewed in the left anterior oblique position, but in the anterior posterior position their shapes are usually quite different In infancy a single ventricle with a rudimentary outlet chamber usually shows fullness of the pulmonary conus

As the infant grows to childhood and the diaphragm descends, the heart comes to occupy a more vertical position and the fullness caused by the rudimentary outflow chamber disappears The differential diagnosis is especially difficult when the electrocardiogram shows a left axis deviation, as in the case reported by Neill and Brink [2] The unipolar precordial leads are usually of aid in the differentiation of the two conditions In tricuspid atresia there is evidence from birth of left ventricular dominance in V_1, whereas in a single ventricle, although there may be a prominent R in V_5 or V_6, the left ventricular pattern is seldom pronounced in V_1

A single ventricle may occur in combination with pulmonary stenosis In

have become sufficiently enlarged to be of functional importance in the maintenance of the pulmonary circulation, or the pulmonary artery persists as a functioning tube which arises from a small right ventricle, which in turn receives its blood from the left ventricle Under such circumstances the condition may be compatible with life for a number of years Nevertheless, most of these patients develop severe polycythemia and are extremely incapacitated A Blalock Taussig operation may greatly improve the prognosis

SUMMARY

Tricuspid atresia is always associated with defective development of the right ventricle Usually this chamber is absent or exists as a diminutive non functioning chamber In addition, there is generally pulmonary atresia Occasionally, when there is tricuspid atresia, the primitive outflow tract develops into a rudimentary outflow chamber which receives blood from the left ventricle Under such circumstances the pulmonary artery may arise in the normal fashion Nevertheless, there is difficulty in the direction of blood to the lungs, hence there is real or functional pulmonary stenosis except in the rare instances in which there is a complete transposition of the great vessels (see Section B)

When the tricuspid valve is atretic the blood from the right auricle is shunted into the left auricle and thence to the left ventricle The mixture of venous and arterial blood reaches the lungs either through a stenosed pulmonary orifice or by way of the ductus arteriosus Consequently there is both a mixture of venous and arterial blood in the left auricle and a reduction in the pulmonary blood flow Hence there is persistent cyanosis

Difficulty in feeding and failure to gain are common complaints

Attacks of paroxysmal dyspnea are common and may be fatal

Squatting is common in patients who survive to childhood

The liver may or may not be enlarged and there may or may not be pulsations palpable at the margin of the liver The occurrence of a pulsating liver which is not engorged is indicative of tricuspid atresia and only a small opening in the auricular septum

The heart is but slightly enlarged The second sound at the base is pure Murmurs may or may not be present.

The diagnosis is suggested by the shape of the heart as seen in the x ray or by fluoroscopy In the anterior posterior view the shadow cast by the pulmonary conus is absent In the left anterior-oblique position the right ventricle does not project forward toward the anterior chest wall beyond the margin of the aorta and the pulmonary window is abnormally clear As the patient grows, the heart

This is notably true when there is only a small defect in the auricular septum or when the foramen ovale is covered by a valve Under such circumstances the increased volume of blood returned from the lungs will close the foramen ovale and the patient will develop severe right sided heart failure Therefore, if the defect in the auricular septum is small, it may be necessary to enlarge the auricular defect as well as to perform the Blalock-Taussig operation

Although some patients have been greatly helped by the combined procedure, over a period of years results have not been as good as those for patients with a tetralogy of Fallot, some patients with tricuspid atresia develop progressive cardiac enlargement and die of cardiac failure within six to eight months after operation Inasmuch as the tricuspid orifice is atretic and the right ventricle is abnormally small, there is virtually no hope of corrective surgery

Glenn[3] has proposed an end to-end anastomosis of the superior vena cava to the right pulmonary artery in order to direct venous blood to the lungs and by pass the right ventricle The operation has proved beneficial to some patients but carries the risk of cerebral edema The long time results cannot as yet be evaluated but decrease in the size of the anastomosis would increase the cerebral pressure

In infants failure to gain or the occurrence of attacks of paroxysmal dyspnea is usually an indication for operation Whenever possible it is advisable to determine the oxygen saturation of the arterial blood If the oxygen saturation of the arterial blood at rest is 20 per cent or less, the danger of death from anoxemia is so great that it is justifiable to assume the risk involved in operation

In older children, however, it is important to remember that after operation there is far greater danger of development of cardiac failure for these patients than for patients with a tetralogy of Fallot Therefore, only if the child is severely incapacitated is operation justified Nevertheless, some children have been greatly helped for many years One of the author s patients did well for eight years and then cyanosis recurred, now he has again been improved by a second anastomosis performed ten years after his first operation

PROGNOSIS

When tricuspid atresia is combined with a non functioning right ventricle and pulmonary atresia, the prognosis is extremely poor As the ductus arteriosus undergoes obliteration the condition usually becomes incompatible with life, the duration of life is usually but three to four months Infants, however, have been known to live for fifteen months When a patient survives for more than two years, either the ductus arteriosus has remained patent, the bronchial arteries

circumstances the right ventricle usually develops as a small chamber which receives its blood from the *left ventricle* The aorta arises from the right ventricle and the pulmonary artery from the left ventricle Inasmuch as the right ventricle lacks its normal inflow tract, some opening *in the auricular septum is inevitable*, either the foramen ovale remains patent or there is a *gross defect* in the auricular septum Furthermore, since no blood *enters the right ventricle from the right* auricle, the only way for blood to reach the right ventricle is from the left ventricle, there must also be a defect in the ventricular septum *Indeed, if there is no* defect in the ventricular septum, the right ventricle is a blind sac and the transposed aorta is markedly hypoplastic and atretic at its base

When the defect in the ventricular septum is small, the aorta, which arises from the right ventricle, may be hypoplastic, as shown in Figure VIII–16 When, however, the defect in the ventricular septum is relatively large, the aorta may be of normal size

The pulmonary artery also may vary in size, it may be of normal size with or without pulmonary stenosis, or it may be abnormally small Figure VIII–17 shows a heart in which there was tricuspid atresia, a relatively large defect in the ventricular septum with an aorta of normal size arising from the right ventricle, and an abnormally small pulmonary artery arising from the left ventricle

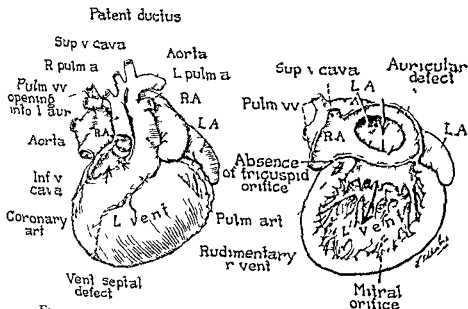


FIGURE VIII–16 Tricuspid atresia combined with complete transposition of the great vessels and a hypoplastic aorta arising from a rudimentary right ventricle Infant

comes to occupy a more vertical position and its contour becomes similar to that of a tetralogy of Fallot In the anterior posterior view the contour is, however, somewhat square and in the left anterior oblique position the anterior border of the heart and the aorta form an almost vertical line

The electrocardiogram usually shows a left axis deviation and the unipolar precordial leads uniformly show evidence of left ventricular dominance in V_1 This finding is so constant that its presence should always suggest a serious abnormality of the tricuspid valve and of the right ventricle

Laboratory studies show evidence of polycythemia The circulation time is abnormally short The oxygen saturation of the arterial blood is reduced and falls still further with exercise Cardiac catheterization gives little information, as it is only possible to pass the catheter from the right auricle to the left auricle Angiocardiography is not of great diagnostic aid

The diagnosis is based on the clinical, fluoroscopic, and the all important electrocardiographic findings

The malformation requires differentiation from a tetralogy of Fallot and from a single ventricle with pulmonary stenosis, and occasionally from a truncus arteriosus with markedly reduced pulmonary blood flow and also from defective development of the right ventricle without tricuspid atresia

Treatment is directed to increase the pulmonary blood flow, and either a Blalock anastomosis or a Potts procedure will help If the defect in the auricular septum is small, it is necessary to combine a systemic pulmonary anastomosis with enlargement of the defect in the auricular septum The long time results are not as good as for a patient with a tetralogy of Fallot, because after operation a number of patients develop progressive cardiac enlargement and intractable cardiac failure Glenn s operation may be of real benefit.

The prognosis in patients with a non functioning right ventricle is extremely poor If the right ventricle receives some blood from the left ventricle, the patient may survive for a number of years Such patients are usually severely incapacitated The prognosis may be greatly improved by operation

B *Tricuspid Atresia Combined with Complete Transposition of the Great Vessels*

NATURE OF THE MALFORMATION

The essential feature of the malformation is the combination of a complete transposition of the great vessels and atresia of the tricuspid valve Under such

circumstances the right ventricle usually develops as a small chamber which receives its blood from the left ventricle The aorta arises from *the right ventricle* and the pulmonary artery from the left ventricle Inasmuch as the right ventricle lacks its normal inflow tract, some opening in the auricular septum is *inevitable*, either the foramen ovale remains patent or there is a gross defect in the auricular septum Furthermore, since no blood enters the right ventricle from *the right* auricle, the only way for blood to reach the right ventricle is from the left ventricle, there must also be a defect in the ventricular septum *Indeed, if there is no* defect in the ventricular septum, the right ventricle is a blind sac and the transposed aorta is markedly hypoplastic and atretic at its base

When the defect in the ventricular septum is small, the aorta, which arises from the right ventricle, may be hypoplastic, as shown in Figure VIII-16 When, however, the defect in the ventricular septum is relatively large, the aorta may be of normal size

The pulmonary artery also may vary in size, it may be of normal size with or without pulmonary stenosis, or it may be abnormally small Figure VIII-17 shows a heart in which there was tricuspid atresia, a relatively large defect in the ventricular septum with an aorta of normal size arising from the right ventricle, and an abnormally small pulmonary artery arising from the left ventricle

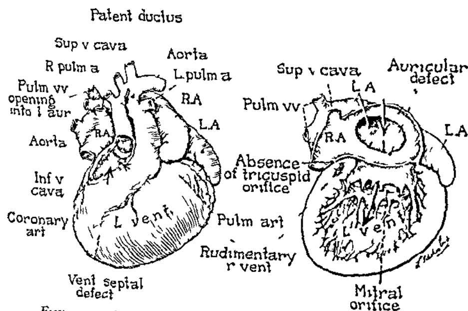


FIGURE VIII-16 Tricuspid atresia combined with complete transposition of the great vessels and a hypoplastic aorta arising from a rudimentary right ventricle Infant

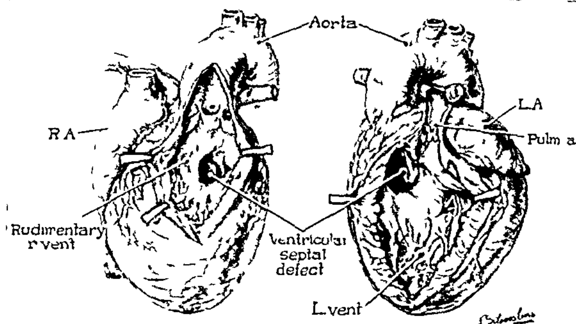


Figure viii-17 Tricuspid atresia combined with complete transposition of the great vessels and a small pulmonary artery arising from the left ventricle (same patient as in Figure viii-19) Child

COURSE OF THE CIRCULATION

During fetal life the circulation is altered both by the absence of the tricuspid orifice and by the abnormal position of the aorta All the blood from the right auricle must flow to the left auricle and thence to the left ventricle If there is aortic atresia, all the blood from the left ventricle is pumped out through the pulmonary artery to the lungs and through the ductus arteriosus to the body When, however, the aorta comes off the right ventricle, some blood is pumped out through the pulmonary artery to the lungs and some through the septal defect into the right ventricle and thence into the aorta The blood which flows to the lungs is returned in the normal manner to the left auricle and that which flows to the body is returned by the superior vena cava and inferior vena cava to the right auricle, as shown in Figure viii-18

After birth the course of the circulation remains essentially the same The atresia of the tricuspid orifice causes all the blood from the right auricle to flow into the left auricle, where it meets the blood returned from the lungs This mixture of oxygenated and venous blood flows into the left ventricle If there is aortic atresia, all the blood from the left ventricle is pumped out into the pulmonary artery Thence part of the blood flows to the lungs and part through the

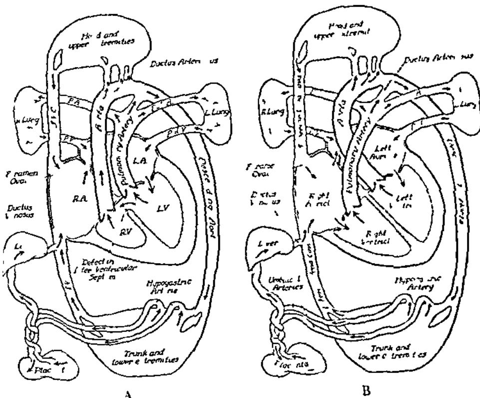


FIGURE VIII–18 Fetal circulation (A) Tricuspid atresia and defective development of the right ventricle combined with complete transposition of the great vessels and a high ventricular septal defect and (B) normal heart

ductus arteriosus to the aorta. The course of the circulation is shown in Diagram VIII–4 When the aorta arises from the right ventricle and receives its blood from the left ventricle, part of the blood from the left ventricle is pumped into the pulmonary artery and part is pumped through the ventricular septal defect to the right ventricle and thence into the aorta The blood which is pumped into the pulmonary artery goes to the lungs, where it is oxygenated, and returned by the pulmonary veins to the left auricle, the blood which is pumped from the right ventricle into the aorta goes to the body and is returned by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again (see Diagram VIII–5)

PHYSIOLOGY OF THE MALFORMATION

In this type of transposition the left auricle receives an admixture of venous blood from the right auricle and oxygenated blood from the lungs, this mixture

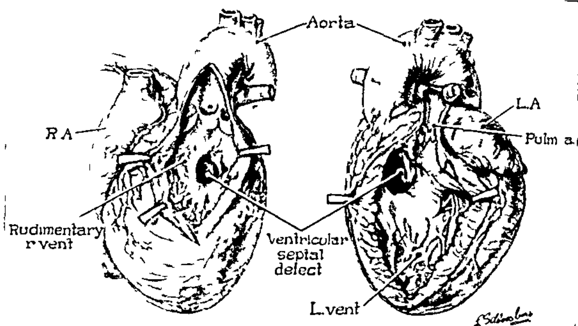


FIGURE VIII-17 Tricuspid atresia combined with complete transposition of the great vessels and a small pulmonary artery arising from the left ventricle (same patient as in Figure VIII-19) Child

COURSE OF THE CIRCULATION

During fetal life the circulation is altered both by the absence of the tricuspid orifice and by the abnormal position of the aorta All the blood from the right auricle must flow to the left auricle and thence to the left ventricle If there is aortic atresia, all the blood from the left ventricle is pumped out through the pulmonary artery to the lungs and through the ductus arteriosus to the body When, however, the aorta comes off the right ventricle, some blood is pumped out through the pulmonary artery to the lungs and some through the septal defect into the right ventricle and thence into the aorta The blood which flows to the lungs is returned in the normal manner to the left auricle and that which flows to the body is returned by the superior vena cava and inferior vena cava to the right auricle, as shown in Figure VIII-18

After birth the course of the circulation remains essentially the same The atresia of the tricuspid orifice causes all the blood from the right auricle to flow into the left auricle, where it meets the blood returned from the lungs This mixture of oxygenated and venous blood flows into the left ventricle If there is aortic atresia, all the blood from the left ventricle is pumped out into the pulmonary artery Thence part of the blood flows to the lungs and part through the

DIAGRAM VIII-4

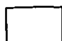
Tricuspid atresia and a non functioning right ventricle combined with complete transposition of the great vessels an auricular septal defect atresia of the aorta at its base and a patent ductus arteriosus

This malformation is a combination of two anomalies namely, (1) a non functioning right ventricle and (2) a complete transposition of the great vessels combined with atresia of the aorta at its base

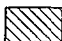
Owing to the tricuspid atresia the blood which enters the right auricle cannot leave by its normal pathway and must escape through a defect in the auricular septum into the left auricle In the left auricle there is complete admixture of the venous blood from the systemic circulation with the oxygenated blood returned by the lungs to the left auricle This mixture of oxygenated and venous blood passes into the left ventricle and is pumped out through the transposed pulmonary artery to the lungs Inasmuch as the aorta is atretic at its base the only way for the blood to reach the systemic circulation is through the ductus arteriosus The blood which is directed to the body is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

Clinical diagnosis Cyanosis is always intense The circulation to the body is poor Inasmuch as the blood is pumped from the left ventricle to the lungs and through the ductus arteriosus to the body there is systemic pressure in the pulmonary artery and hence there is pulmonary hypertension Moreover a large volume of blood is pumped to the lungs pulmonary congestion occurs early and may be extreme The circulation may be so poor that the duration of life is too short to produce cardiac hypertrophy The heart is normal in size The x ray shows an absence of the fullness of the pulmonary conus in the anterior posterior position and a small right ventricle in the left anterior-oblique position The electrocardiogram shows a left axis deviation and evidence of combined hypertrophy and left ventricular dominance

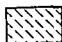
Diagram VIII–4



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

Diagram VIII–5

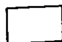
Tricuspid atresia and complete transposition of the great vessels an auricular septal defect and a ventricular septal defect

This malformation combines a tricuspid atresia and a complete transposition of the great vessels The occurrence of the tricuspid atresia renders inevitable a defect in the auricular septum The origin of the aorta from the right ventricle which lacks its normal inflow tract, renders inevitable a defect in the ventricular septum

Because of the tricuspid atresia the blood from the right auricle cannot escape by way of its normal channel it can escape only through the defect in the auricular septum into the left auricle. There it mixes with the blood which enters the left auricle from the pulmonary veins This admixture of venous and arterial blood flows into the left ventricle Part of the blood from the left ventricle is pumped out by way of the pulmonary artery to the lungs and the oxygenated blood is again returned to the left auricle and the left ventricle Part of the blood is forced through the ventricular septal defect into the right ventricle and thence it is pumped out by way of the aorta to the systemic circulation. The blood from the head and the upper extremities is returned in the normal fashion by the superior vena cava and that from the lower extremities is returned by the inferior vena cava to the right auricle. There the cycle starts again

Clinical diagnosis The complete admixture of venous and arterial blood, combined with the difficulty in the direction of the blood to the systemic circulation causes intense cyanosis The tricuspid atresia causes the right ventricle to be smaller than the left ventricle. Consequently the electrocardiogram shows a left axis deviation and evidence of combined hypertrophy and left ventricular preponderance The complete transposition of the great vessels leads to progressive cardiac enlargement and cardiac failure The fact that the pulmonary artery is of normal size and arises from the left ventricle means that the lungs receive blood under systemic pressure The pulmonary circulation however is adequate or excessive and congestion of the lungs is common In brief, this malformation shows a combination of the features characteristic of a complete transposition of the great vessels and those which result from the tricuspid atresia and defective development of the right ventricle

Diagram VIII–5



Arterial blood (fully saturated)



Small admixture of venous blood
No visible cyanosis



Venous and arterial blood
Cyanosis visible



Venous blood

Diagram VIII–5

Tricuspid atresia and complete transposition of the great vessels an auricular septal defect and a ventricular septal defect

This malformation combines a tricuspid atresia and a complete transposition of the great vessels The occurrence of the tricuspid atresia renders inevitable a defect in the auricular septum The origin of the aorta from the right ventricle, which lacks its normal inflow tract renders inevitable a defect in the ventricular septum

Because of the tricuspid atresia the blood from the right auricle cannot escape by way of its normal channel, it can escape only through the defect in the auricular septum into the left auricle. There it mixes with the blood which enters the left auricle from the pulmonary veins This admixture of venous and arterial blood flows into the left ventricle. Part of the blood from the left ventricle is pumped out by way of the pulmonary artery to the lungs and the oxygenated blood is again returned to the left auricle and the left ventricle Part of the blood is forced through the ventricular septal defect into the right ventricle and thence it is pumped out by way of the aorta to the systemic circulation. The blood from the head and the upper extremities is returned in the normal fashion by the superior vena cava and that from the lower extremities is returned by the inferior vena cava to the right auricle. There the cycle starts again

Clinical diagnosis The complete admixture of venous and arterial blood, combined with the difficulty in the direction of the blood to the systemic circulation, causes intense cyanosis The tricuspid atresia causes the right ventricle to be smaller than the left ventricle Consequently the electrocardiogram shows a left axis deviation and evidence of combined hypertrophy and left ventricular preponderance The complete transposition of the great vessels leads to progressive cardiac enlargement and cardiac failure The fact that the pulmonary artery is of normal size and arises from the left ventricle means that the lungs receive blood under systemic pressure The pulmonary circulation however is adequate or excessive and congestion of the lungs is common In brief this malformation shows a combination of the features characteristic of a complete transposition of the great vessels and those which result from the tricuspid atresia and defective development of the right ventricle

of venous and arterial blood flows into the left ventricle Consequently the same admixture of venous and arterial blood is pumped into the aorta and out through the pulmonary artery to the lungs If there is no stenosis of the pulmonary orifice, the total circulation to the lungs is normal or excessive but the effective flow is reduced Furthermore, the origin of the pulmonary artery from the left ventricle means that blood is pumped into the pulmonary artery under the same pressure as it is pumped into the aorta Consequently, unless there is stenosis of the pulmonary valve, there is always pulmonary hypertension The adequacy of the systemic circulation depends upon the size of the ventricular defect When the defect is minute or the aorta is atretic at its base, the condition is physiologically and clinically similar to that of aortic atresia and the duration of life is extremely short (see Chapter XIII, Section A)

The larger the defect the more readily is blood directed to the aorta hence the duration of life is longer Nevertheless, the condition usually leads to progressive cardiac enlargement and cardiac failure at an early age

If there is pulmonary stenosis, more blood is forced through the septal defect to the right ventricle, the systemic circulation is thereby increased and less blood goes to the lungs for oxygenation Under such circumstances, although the lungs are protected from the high pressure in the left ventricle, the effective flow may be greatly reduced and the oxygen saturation of the arterial blood is proportionally low

CLINICAL FINDINGS

Cyanosis dates from birth and is of uniform distribution It is, however, not necessarily intense

Respirations are rapid When there is pulmonary atresia or extreme pulmonary stenosis, the infant may suffer from attacks of paroxysmal dyspnea Such attacks are absent if the pulmonary artery is small but the pulmonary pressure is high

Pulmonary congestion occurs when the pulmonary artery is of normal size If the aorta is atretic, pulmonary congestion occurs early and remains severe throughout the infant's brief life The occurrence of pulmonary stenosis or an abnormally small pulmonary artery protects the lungs and renders the condition more compatible with life

The exercise tolerance of the child varies but it is not severely limited unless there is real difficulty in the direction of blood to the lungs

The liver is usually enlarged and may extend to the umbilicus

Edema is a late manifestation

CARDIAC FINDINGS

The size of the heart varies with the size of the aorta and the pulmonary artery When either of the great vessels is atretic, the duration of life is so short that the heart remains small

When both great vessels are of functional importance, the duration of life is longer Indeed, when the aorta is of normal size and the pulmonary artery is small or moderately stenosed, the condition may be compatible with life for a number of years The heart becomes moderately enlarged

The second heart sound at the base is accentuated as the aorta is displaced anteriorly and to the left Reduplication of the second heart sound indicates that both great vessels are of relatively normal size *Murmurs* are of no great diagnostic aid Usually there is a *systolic* murmur A *gallop rhythm* may or may not be present

Progressive cardiac enlargement is the rule when both great vessels are of normal size Consequently the condition leads to cardiac failure at an early age

Cardiac failure is manifested by congestion of the lungs, engorgement of the liver, and edema of the extremities There may even be ascites

X RAY AND FLUOROSCOPIC FINDINGS

The contour of the heart varies with the relative size of the great vessels, the age of the patient, and the duration of life

When the pulmonary artery is atretic the lungs are excessively clear and the contour of the heart is similar to that in other cases of a non functioning right ventricle, as shown in Figures VIII–6 and 8

When the aorta is atretic at its base and the pulmonary artery is of normal size, although the contour of the heart is similar to that which occurs with pulmonary atresia and a non functioning right ventricle, the lungs are not unduly clear but duration of life may be too short for hilar shadows to become conspicuous

When the aorta is of normal size and the pulmonary artery is abnormally small the contour of the heart is somewhat square This contour is accentuated when the aorta arches to the right and displaces the superior vena cava to the right (as shown in Figure VIII–19) and under such circumstances examination in the left anterior-oblique position will show that the right ventricle is small The lungs are clear The pulmonary window is also clear because the posteriorly placed pulmonary artery courses to the lungs at an abnormally low level The structure of the heart in this combination of anomalies is illustrated in Figure VIII–17 and the x ray of the case is shown in Figure VIII–19

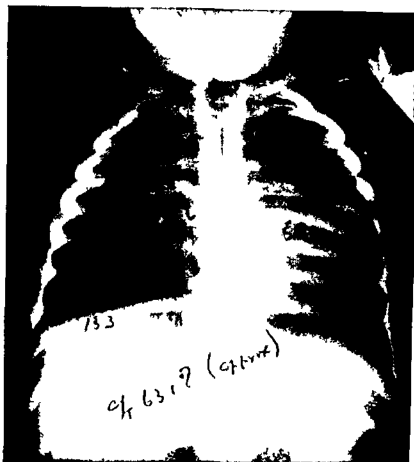


FIGURE VIII-19 Tricuspid atresia combined with complete transposition of the great vessels and a small pulmonary artery (same patient as in Figure VIII-17) Child

When both great vessels are of normal size, the heart undergoes progressive cardiac enlargement, as it does in other cases of *complete transposition* of the great vessels, both ventricles become enlarged and the right auricle may be dilated There is a concave curve at the base of the heart to the left of the sternum and congestion in the lungs, as shown in Figure VIII-20 Examination of the patient in the left anterior oblique position shows great enlargement posteriorly and little or no enlargement anteriorly

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads usually show a left axis deviation and almost invariably the unipolar precordial leads will show evidence of left ventricular dominance, which increases as the patient grows older

SPECIAL TESTS

The red blood cell count, the amount of available hemoglobin and *the hematocrit reading* are always increased

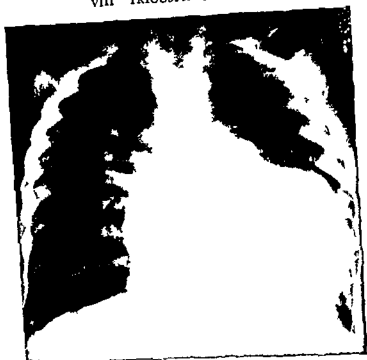


FIGURE VIII–20 Tricuspid atresia combined with complete transposition of the great vessels and a large pulmonary artery Child

The oxygen saturation of the arterial blood is always reduced

Cardiac catheterization is usually of no great diagnostic aid, as it is frequently impossible to pass the catheter beyond the right auricle

Angiocardiography shows prompt opacification of the left auricle immediately after the right auricle is filled, and before either ventricle is delineated When both great vessels are present, they are visualized simultaneously In the lateral film the aorta is seen to lie anterior to the pulmonary artery

DIAGNOSIS

The diagnosis is based upon the finding of a left axis deviation and evidence of left ventricular dominance in the electrocardiogram of a cyanotic patient with a heart which has a concave curve at its base to the left of the sternum When this finding is combined with increased vascularity of the lungs, it is clear evidence that the pulmonary artery is transposed and is of normal size

When the pulmonary artery is small the diagnosis is difficult, as the condition simulates a non functioning right ventricle without transposition of the great vessels The absence of paroxysmal dyspnea and of squatting suggests that the tricuspid atresia is combined with a transposition of the great vessels

When a complete transposition of the great vessels combined with atresia of

the aorta at its base is associated with tricuspid atresia and a non functioning right ventricle, the course of the circulation is similar to that of other types of aortic atresia and the contour of the heart and the electrocardiographic findings are similar to those of a non functioning right ventricle

DIFFERENTIAL DIAGNOSIS

The condition requires differentiation from tricuspid atresia without transposition of the great vessels, from a single ventricle, and from other types of complete transposition of the great vessels

Tricuspid atresia without transposition of the great vessels is always associated with reduced pulmonary blood flow As previously mentioned, the contour of the heart in this malformation closely resembles that which is found in tricuspid atresia when the great vessels are transposed Angiocardiography may be necessary to determine which of the great vessels lies anterior to the other

A single ventricle may be confused with tricuspid atresia and transposition of the great vessels Usually the electrocardiogram differentiates the two conditions If the electrocardiogram shows a left axis deviation and evidence of left ventricular hypertrophy, cardiac catheterization may be necessary to determine whether the left auricle or the common ventricle is entered from the right auricle

Other types of complete transposition of the great vessels show electrocardiographic evidence of a right axis deviation and right ventricular hypertrophy

TREATMENT

Medical treatment is palliative, surgical treatment is difficult and often impossible

When either of the great vessels is atretic the condition of the infant is too precarious for surgery to be attempted

When both great vessels are of functional importance there are three major considerations to be taken into account (1) the adequacy of the pulmonary blood flow, (2) the size of the opening between the two auricles, and (3) the size of the ventricular defect

When there is pulmonary stenosis, the circulation to the lungs may be increased by a systemic pulmonary anastomosis, but if such is attempted due consideration must be given to the size of the auricular defect If the only communication between the two auricles is the patency of the foramen ovale, the increased circulation to the lungs will close the valve and precipitate right sided

cardiac failure, therefore it may also be necessary to create an auricular defect The heart, however, may not be able to adjust to the altered circulation

In some instances the ventricular septal defect is so small that it is difficult for the left ventricle to expel the blood which it receives from the lungs The creation of a ventricular septal defect is far more difficult than that of an auricular defect At best it would only change the malformation to that of a single ventricle

PROGNOSIS

The prognosis is poor The duration of life depends upon the size of the great vessels and the size of the intracardiac shunts The condition frequently leads to progressive cardiac enlargement and cardiac failure Most infants with this malformation die in the first year of life, a few may live to childhood

SUMMARY

Tricuspid atresia may occur in combination with a complete transposition of the great vessels Under such circumstances the right ventricle, from which the aorta arises, is of functional importance and receives its blood from the left ventricle through a defect in the ventricular septum The pulmonary artery, which arises from the left ventricle, may be normal in size, abnormally large, or stenotic Unless there is pulmonary stenosis, there is always pulmonary hypertension Inasmuch as the aorta arises from the right ventricle, which receives its blood from the left ventricle, there may be difficulty in the establishment of the systemic circulation

Cyanosis dates from birth and is usually intense

The exercise tolerance of patients with this condition varies with the severity of the pulmonary stenosis and with the adequacy of the systemic circulation

The heart tends to undergo progressive enlargement The second sound over the pulmonary area is accentuated, murmurs are variable The x ray shows a somewhat square contour with a concave curve at the base of the heart to the left of the sternum The vascularity of the lung fields varies with the size of the pulmonary orifice

The condition requires differentiation from a single ventricle combined with a transposition of the great vessels, and from other types of tricuspid atresia and also other types of complete transposition of the great vessels

Treatment is usually unsatisfactory The structure of the heart is so grossly abnormal that there is little chance of surgical relief Only if the patient suffers

from pulmonary stenosis and lack of adequate pulmonary blood flow is there any hope that a systemic pulmonary anastomosis will help, and even then it may be necessary to combine this procedure with the creation of an auricular septal defect

References

1 Taussig, H B The clinical and pathological findings in congenital malformations of the heart due to defective development of the right ventricle associated with tricuspid atresia or hypoplasia Bull Johns Hopkins Hosp 59 435-445, 1936

2 Neill, C A and A J Brink Left axis deviation in tricuspid atresia and single ventricle, *the electrocardiogram in 36 autopsied cases Circulation 12 612-619 1955*

3 Glenn W W L Circulatory by pass of the right side of the heart iv Shunt between superior vena cava and distal right pulmonary artery report of clinical application New England J of Med 259 117-120, 1958

CHAPTER IX

DEFECTIVE DEVELOPMENT OF THE RIGHT VENTRICLE

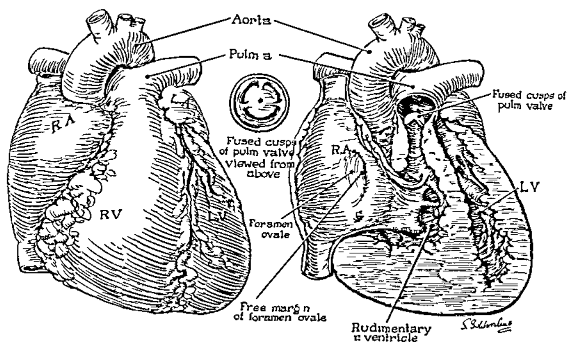
DEFECTIVE development of the right ventricle may occur as an isolated malformation with an intact ventricular septum and without tricuspid atresia There are at least two types of abnormality in the development of the right ventricle associated with a normal tricuspid orifice The more common type is the one in which the right ventricle is abnormally small but is of functional importance in the direction of blood to the lungs In such instances the pulmonary artery is abnormally small and usually its orifice is stenotic In rare instances the pulmonary stenosis is so extreme that there is virtual pulmonary atresia The malformation differs from the usual pulmonary stenosis with an intact ventricular septum in that the right ventricle is so small that it cannot pump its full load and is unable to dilate to meet the demands of the body Clinically the condition more closely resembles a tetralogy of Fallot with severe pulmonary stenosis or atresia than it does a pulmonary stenosis with an intact ventricular septum This anomaly is presented in Section A

The other type occurs when the defect in the development of the right ventricle is of such a nature that the right ventricle is a large chamber with a thin, flabby wall The wall is so poorly developed that the right ventricle is an ineffective pump The electrocardiogram usually shows a left axis deviation This condition is discussed in Section B

A *Defective Development of the Right Ventricle with an Intact Ventricular Septum*

NATURE OF THE MALFORMATION

In this malformation the right ventricle fails to expand and grow normally it remains a small chamber A small tricuspid valve opens into it in the normal manner The ventricular septum is intact and a small pulmonary artery arises from this chamber Usually there is a cap like stenosis of the pulmonary valve Figure IX–1 illustrates a heart of this type The stenosis may be so extreme that there is a functional pulmonary atresia Sometimes the infundibular tract is extremely narrow but leads to a diminutive pulmonary valve with tiny but nor



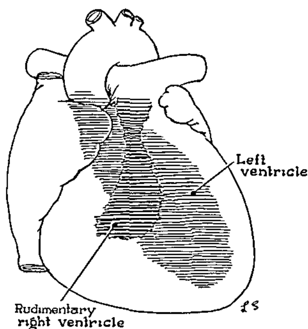


FIGURE IX-1 Defective development of the right ventricle with valvular pulmonary stenosis and an intact ventricular septum (Case IX-1) Child

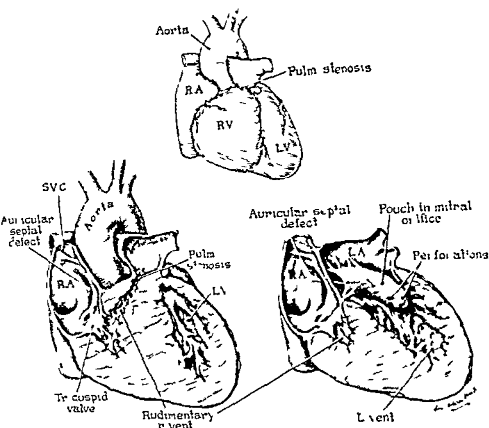


Figure IX–2 Defective development of the right ventricle with extreme infundibular stenosis

Note also the pouch extending from the left auricle into the left ventricle

mally formed cusps, as shown in Figure IX–2 In the latter instance the right ventricle was abnormally small, with marked narrowing of the outflow tract and a small pulmonary artery, and in addition there was an extraordinary membranous pouch which extended from the foramen ovale into the left auricle and partially obstructed the mitral orifice Consequently the infant suffered not only from an abnormally small right ventricle and pulmonary stenosis but also from mitral stenosis Little wonder that in this instance life was of such short duration that no studies were obtained

As in all types of malformations there are all grades of severity The right ventricle is always abnormally small but, if the pulmonary stenosis is not ex

tremely severe, the patient may live to childhood, whereas, if the stenosis is extremely severe or if there is pulmonary atresia, the condition leads to progressive cardiac enlargement and cardiac failure in early infancy If extreme pulmonary stenosis is combined with a diminutive right ventricle, the condition is functionally similar to a tricuspid atresia with a non functioning right ventricle

COURSE OF THE CIRCULATION

The blood from the right auricle is directed to the right ventricle in the normal manner and is pumped out through the stenosed pulmonary artery to the lungs, where it is oxygenated and returned in the normal manner to the left auricle The small size of the right ventricle renders it impossible for the right auricle to empty in the normal manner Hence the pressure in the right auricle is abnormally high, because of this the foramen ovale is held open and some blood is shunted from the right auricle into the left auricle There the venous blood from the right auricle meets the oxygenated blood returned from the lungs to the left auricle This admixture of venous and oxygenated blood flows into the left ventricle and is pumped out through the aorta to the systemic circulation and returned by the superior and inferior venae cavae to the right auricle There the cycle starts again, as shown in Diagram IX–1 When there is pulmonary atresia, the course of the circulation is essentially the same as that shown in Diagram VIII–1

PHYSIOLOGY OF THE MALFORMATION

The small size of the right ventricle and the pulmonary obstruction increase the pressure in the right ventricle, which in turn raises the pressure in the right auricle The high pressure in the right auricle holds the foramen ovale open and a right to-left shunt is established at the auricular level The pulmonary stenosis protects the lungs, so that the pulmonary vascular bed is normal

CLINICAL FINDINGS

Cyanosis is usually present at birth or shortly thereafter and increases as the ductus arteriosus undergoes obliteration

Clubbing of the extremities occurs early

Polycythemia develops at an early age The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are all abnormally high If the duration of life permits, polycythemia may become extreme

Dyspnea is common but attacks of paroxysmal dyspnea rarely occur

Growth and *development* are retarded Infants with this malformation usually gain weight extremely slowly

Exercise tolerance is limited but the child seldom squats when tired When short of breath, he simply stops to rest As the child grows, he gradually becomes progressively more incapacitated This is in striking contrast to the child with a tetralogy of Fallot, who usually improves between two and five years of age

The liver is often enlarged and may be huge, frequently there are *pulsations* palpable at the margin of the liver

Edema and *ascites* are late manifestations

CARDIAC FINDINGS

The size of the heart varies with the severity of the abnormality The heart is always somewhat enlarged The condition leads to progressive cardiac enlargement The rate of enlargement varies with the size of the right ventricle and the severity of the pulmonary stenosis Thus there may be only slight enlargement at six years of age, as shown in Figure IX–3, or there may be tremendous enlargement at four months of age, as shown in Figure IX–4 The *second sound* to the left of the sternum is weak or absent A *systolic murmur* may or may not be audible

The malformation causes great cardiac enlargement and terminal cardiac failure with engorgement of the liver and edema of the extremities and even ascites The lungs, however, usually remain clear

X RAY AND FLUOROSCOPIC FINDINGS

The heart is usually enlarged and may be huge In the anterior posterior position there is a narrow pedicle at the base of the heart and the upper margin of the shadow to the left of the sternum has a concave curve The right auricle is slightly enlarged and the vascular markings are decreased, as shown in Figure IX–3

In the left anterior-oblique position there is no enlargement of the right ventricle but the left ventricle extends abnormally far posteriorly Owing to the pulmonary stenosis the pulmonary window is clear

If the pulmonary stenosis is extreme, the right auricle and the left ventricle are enlarged, the heart lies horizontally upon the diaphragm, and the lungs are excessively clear as shown in Figure IX–4 Under such circumstances, in the left anterior-oblique view, the enlargement of the right auricle *may extend so far an*teriorly as to suggest great right ventricular enlargement and the ventricular

tremely severe, the patient may live to childhood, whereas, if the stenosis is extremely severe or if there is pulmonary atresia, the condition leads to progressive cardiac enlargement and cardiac failure in early infancy If extreme pulmonary stenosis is combined with a diminutive right ventricle, the condition is functionally similar to a tricuspid atresia with a non functioning right ventricle

COURSE OF THE CIRCULATION

The blood from the right auricle is directed to the right ventricle in the normal manner and is pumped out through the stenosed pulmonary artery to the lungs, where it is oxygenated and returned in the normal manner to the left auricle The small size of the right ventricle renders it impossible for the right auricle to empty in the normal manner Hence the pressure in the right auricle is abnormally high, because of this the foramen ovale is held open and some blood is shunted from the right auricle into the left auricle There the venous blood from the right auricle meets the oxygenated blood returned from the lungs to the left auricle This admixture of venous and oxygenated blood flows into the left ventricle and is pumped out through the aorta to the systemic circulation and returned by the superior and inferior venae cavae to the right auricle There the cycle starts again, as shown in Diagram IX–I When there is pulmonary atresia, the course of the circulation is essentially the same as that shown in Diagram VIII–I

PHYSIOLOGY OF THE MALFORMATION

The small size of the right ventricle and the pulmonary obstruction increase the pressure in the right ventricle, which in turn raises the pressure in the right auricle The high pressure in the right auricle holds the foramen ovale open and a right to-left shunt is established at the auricular level The pulmonary stenosis protects the lungs, so that the pulmonary vascular bed is normal

CLINICAL FINDINGS

Cyanosis is usually present at birth or shortly thereafter and increases as the ductus arteriosus undergoes obliteration

Clubbing of the extremities occurs early

Polycythemia develops at an early age The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are all abnormally high If the duration of life permits, polycythemia may become extreme

Dyspnea is common but attacks of paroxysmal dyspnea rarely occur

Diagram IX-1

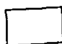
Defective development of the right ventricle with valvular pulmonary stenosis and an intact ventricular septum

The fundamental feature in this malformation is the defective development of the right ventricle, from which a small pulmonary artery arises, in addition there is usually stenosis of the pulmonary valve The ventricular septum is intact The foramen ovale is held open by the high pressure in the right auricle

Part of the blood from the right auricle flows into the small right ventricle and is pumped out through the stenosed pulmonary artery to the lungs, where it is oxygenated and returned to the left auricle in the normal manner Owing to the small size of the right ventricle, it cannot accommodate all the blood which is returned to the right auricle Hence the pressure in the right auricle rises and some blood is forced through the foramen ovale to the left auricle and mixes with the oxygenated blood returned by the pulmonary veins to the left auricle This admixture of arterial and venous blood flows into the left ventricle and is pumped out through the aorta to the systemic circulation It is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

Clinical diagnosis The infant suffers from persistent cyanosis, which dates from birth but does not suffer from attacks of paroxysmal dyspnea, nevertheless the patient's incapacity becomes progressively greater as he grows older The heart is slightly to moderately enlarged with a concave curve at its base. The second sound over the pulmonary area is diminished and the pulmonary vascular markings are reduced The electrocardiogram shows only slight evidence of hypertrophy of the right ventricle in V_1 and an early transition zone to left ventricular dominance

Diagram IX-1


Arterial blood (fully saturated)


Venous and arterial mixed
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood



Left anterior-oblique position



Right anterior-oblique position

FIGURE IX-5 Defective development of the right ventricle with extreme pulmonary stenosis and an intact ventricular septum (same patient as in Figure IX-4) Infant

shadow extends abnormally far posteriorly (see *Figure IX-5*) The pulmonary window is clear In the right anterior-oblique position there is no enlargement of the left auricle

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram gives the clue to the diagnosis There may or may not be a right axis deviation The unipolar precordial leads always reveal less evidence of right ventricular hypertrophy than would be expected from the clinical picture There may be an R wave of moderate height in V_1, but never the evi

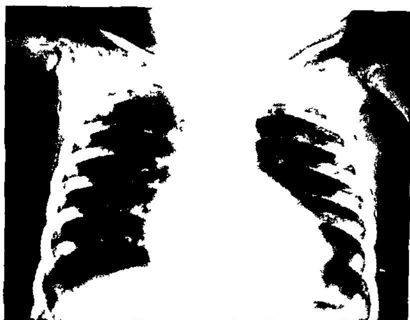


FIGURE IX-3 Defective development of the right ventricle with valvular pulmonary stenosis and an intact ventricular septum (Case IX-1) Child



FIGURE IX-4 Defective development of the right ventricle with extreme pulmonary stenosis and an intact ventricular septum (same patient as in Figures IX-5, 7) Infant



Left anterior oblique position



Right anterior-oblique position

FIGURE IX-5 Defective development of the right ventricle with extreme pulmonary stenosis and an intact ventricular septum (same patient as in Figure IX-4) Infant

shadow extends abnormally far posteriorly (see Figure IX-5) The pulmonary window is clear In the right anterior oblique position there is no enlargement of the left auricle

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram gives the clue to the diagnosis There may or may not be a right axis deviation The unipolar precordial leads always reveal less evidence of right ventricular hypertrophy than would be expected from the clinical picture There may be an R wave of moderate height in V_1, but never the evi

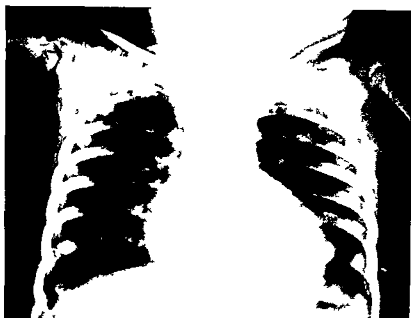


FIGURE IX-3 Defective development of the right ventricle with valvular pulmonary stenosis and an intact ventricular septum (Case IX-1) Child



FIGURE IX-4 Defective development of the right ventricle with extreme pulmonary stenosis and an intact ventricular septum (same patient as in Figures IX-5, 7) Infant

right ventricle are markedly elevated Indeed, the pressure in the right ventricle may be higher than the systemic pressure The aorta cannot be entered from the right ventricle It may not be possible to catheterize the pulmonary artery If the pulmonary artery is entered, the oxygen content will be the same as or lower than that of the right ventricle and the pressure will be low

If the patient is catheterized through the saphenous vein, it is frequently possible to catheterize the left auricle through the foramen ovale, which is held open by the high pressure in the right auricle The oxygen saturation in the left auricle, the left ventricle, and the femoral artery is reduced

Angiocardiography shows a large right auricle in which the dye lingers for some time Usually the dye is seen to pass to the left auricle and the left ventricle as well as to the right ventricle The right ventricle is seen to be small and the pulmonary artery may or may not be visualized, depending on the severity of the pulmonary stenosis The aorta is not opacified until after the left auricle has filled Furthermore, in the lateral view the aorta is seen to arise in its normal position

DIAGNOSIS

The diagnosis is based upon the occurrence of intense cyanosis and increasing incapacity Although cyanosis dates from birth, a history of attacks of paroxysmal dyspnea is seldom obtained and the child rarely squats when tired When the right ventricle is tiny or when the pulmonary artery is atretic at its base, the heart enlarges rapidly and cardiac failure occurs in infancy When the condition is less severe, the malformation may simulate a tetralogy of Fallot but the electrocardiogram shows less evidence of right ventricular hypertrophy

Angiocardiography will show that the right ventricle is abnormally small and that the aorta has no connection with it Cardiac catheterization will reveal that the right ventricular pressure is abnormally high

DIFFERENTIAL DIAGNOSIS

In early infancy the condition requires differentiation from other causes of cardiac enlargement and cardiac failure, notably 'pure' pulmonary stenosis of a severe degree and Ebstein s anomaly of the tricuspid valve, and also from the malformation in which both vessels arise from the right ventricle In older children the condition may be confused with a tetralogy of Fallot, truncus arteriosus with reduced pulmonary blood flow a tricuspid atresia, and possibly a single ventricle with pulmonary stenosis

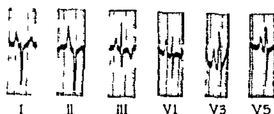


FIGURE IX-6 Defective development of the right ventricle with valvular pulmonary stenosis and an intact ventricular septum (Case IX-1) Child

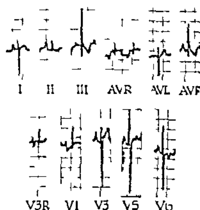


FIGURE IX-7 Defective development of the right ventricle with extreme pulmonary stenosis and an intact ventricular septum (same patient as in Figure IX-4) Infant

dence of extreme right ventricular hypertrophy such as is characteristic of severe pulmonary stenosis with an intact ventricular septum Usually the precordial leads show as much evidence of left ventricular dominance as of right ventricular hypertrophy Thus V_1 may show an S wave of greater height than the R wave, in addition there is frequently an early transition zone to left ventricular dominance The P waves are tall and peaked, definitely of the Himalayan type Indeed, the combination of Himalayan P waves and relatively little evidence of right ventricular dominance in V_1 (see Figures IX-6 and 7) is always suggestive of this malformation

SPECIAL TESTS

The red blood cell count, the amount of available hemoglobin and *the hematocrit reading* are all abnormally high

The oxygen saturation of the arterial blood is reduced and may be extremely low

Cardiac catheterization shows essentially the same oxygen content in the right auricle and the right ventricle The pressures in the right auricle and the

TREATMENT

When the right ventricle is abnormally small and the aorta does not over ride the ventricular septum, a systemic pulmonary anastomosis leads to right sided cardiac failure (see Illustrative Case IX-1)

A valvulotomy is of some benefit but the small size of the right ventricle pre vents that chamber from carrying its full load Indeed, the right ventricle is so small that a transventricular valvulotomy is impossible, hence the operation must be done through the pulmonary artery under direct vision Unfortunately the relief of the pulmonary stenosis does not alter appreciably the size of the right ventricle, which remains abnormally small For this reason, after valvulot omy, no attempt should be made to close the auricular defect, as it acts as an escape valve for the high pressure in the auricle

The larger the right ventricle, the greater is the benefit derived from opera tion. Furthermore, the larger the right ventricle, the longer is the condition com patible with life For this reason the operation is usually of greater benefit to children than to infants

When the right ventricle is extremely small, Glenn's[1] operation to by pass the right ventricle by the anastomosis of the superior vena cava to the right pul monary artery may also be of benefit

PROGNOSIS

The prognosis is poor but varies with the size of the right ventricle When a diminutive right ventricle is combined with pulmonary atresia, the prognosis is extremely poor When the right ventricle is a fair sized chamber, the condition may be compatible with life for a number of years Usually the child becomes progressively more severely incapacitated

Operation either to relieve the pulmonary stenosis or to by pass the right ven tricle alleviates the condition but the abnormality in the size of the right ventricle remains a permanent handicap

SUMMARY

Defective development of the right ventricle with an intact ventricular sep- tum places a great strain on the right side of the heart The condition is almost always associated with pulmonary stenosis, either valvular, infundibular, or both

Cyanosis dates from birth and increases in intensity Dyspnea is marked but the infant does not suffer from attacks of paroxysmal dyspnea

'Pure' pulmonary stenosis with a normal right ventricular cavity always shows far greater electrocardiographic evidence of right ventricular hypertophy in V_1 than does the malformation under discussion, by the time that cardiac failure develops the electrocardiogram usually shows the pattern of so-called right ventricular "strain"

Ebstein's anomaly of the tricuspid valve may be confused with defective development of the right ventricle and an intact ventricular septum in early infancy, because the characteristic electrocardiographic pattern of Ebstein's anomaly is frequently not present in the early months of life In Ebstein's anomaly the engorgement of the liver is not associated with pulsations at its margin and cardiac catheterization reveals a normal pressure in the right ventricle and in the pulmonary artery

The origin of both great vessels from the right ventricle causes early persistent cyanosis and may lead to progressive cardiac enlargement and cardiac failure The electrocardiogram, however, shows clear evidence of right ventricular hypertrophy in the precordial leads Angiocardiography reveals early opacification of the aorta as well as of the pulmonary artery

A tetralogy of Fallot differs from defective development of the right ventricle and an intact ventricular septum in that, when cyanosis appears early, the infant almost invariably suffers from attacks of paroxysmal dyspnea The heart is usually of normal size and the electrocardiogram shows greater evidence of right ventricular hypertrophy in V_1 Angiocardiography will show that the aorta receives blood from the right ventricle

Tricuspid atresia with pulmonary stenosis or atresia differs from the malformation under discussion in that the electrocardiogram shows a left axis deviation and evidence of left ventricular hypertrophy

A truncus arteriosus with reduced pulmonary blood flow causes the heart to assume a totally different contour The condition does not lead to progressive cardiac enlargement and cardiac failure In older children the two malformations are seldom confused

A single ventricle with pulmonary stenosis differs from the malformation under discussion in that, when pulmonary stenosis occurs in combination with a single ventricle, the infant suffers from attacks of paroxysmal dyspnea and the child squats when tired The patient, however, generally improves during early childhood, whereas when the right ventricle is abnormally small the child becomes progressively more incapacitated Furthermore, when there is but a single ventricle, angiocardiography reveals early opacification of the aorta

TREATMENT

When the right ventricle is abnormally small and the aorta does not override the ventricular septum, a systemic pulmonary anastomosis leads to right sided cardiac failure (see Illustrative Case IX–1).

A valvulotomy is of some benefit but the small size of the right ventricle prevents that chamber from carrying its full load. Indeed, the right ventricle is so small that a transventricular valvulotomy is impossible, hence the operation must be done through the pulmonary artery under direct vision. Unfortunately the relief of the pulmonary stenosis *does not alter appreciably* the size of the *right ventricle*, which remains abnormally small. For this reason, after valvulotomy, no attempt should be made to close the auricular defect, as it acts as an escape valve for the high pressure in the auricle.

The larger the right ventricle, the greater is the benefit derived from operation. Furthermore, the larger the right ventricle, the longer *is the condition compatible* with life. For this reason the operation is usually of greater benefit to children than to infants.

When the right ventricle is extremely small, Glenn's[1] operation to by pass the right ventricle by the anastomosis of the superior vena cava to the right pulmonary artery may also be of benefit.

PROGNOSIS

The prognosis is poor but varies with the size of the right ventricle. When a diminutive right ventricle is combined with pulmonary atresia, the prognosis is extremely poor. *When the right ventricle is a fair sized chamber*, the condition may be compatible with life for a number of years. Usually the child becomes progressively more severely incapacitated.

Operation either to relieve the pulmonary *stenosis or to* by pass the right ventricle alleviates the condition but the abnormality in the size of the right ventricle remains a permanent handicap.

SUMMARY

Defective development of the right ventricle with an intact ventricular septum places a great strain on the *right side of the heart*. The condition is almost always associated with pulmonary stenosis either valvular, infundibular, or both.

Cyanosis dates from birth and increases in intensity. Dyspnea is marked but the infant does not suffer from attacks of paroxysmal dyspnea.

The heart undergoes progressive enlargement If the condition is severe, it leads to cardiac failure in early infancy If less severe, the heart enlarges so slowly that the cardiac findings may simulate a tetralogy of Fallot

The electrocardiogram, however, shows less evidence of right ventricular hypertrophy than is usual in a tetralogy of Fallot or in a severe pulmonary stenosis with an intact ventricular septum Nevertheless, the P waves are often of the Himalayan type

Angiocardiography shows a huge right auricle and a small right ventricle, the pulmonary artery may or may not be visualized The aorta is seen to arise in its normal posterior position

Cardiac catheterization reveals high pressure in the right auricle and in the right ventricle, if the pulmonary artery is entered, the pressure will be low

The diagnosis is based on the finding of early deep cyanosis in an infant who does not suffer from paroxysmal dyspnea, who has evidence of right sided cardiac failure, and whose electrocardiogram shows little or no evidence of right ventricular hypertrophy

The condition requires differentiation from 'pure" pulmonary stenosis, Ebstein s anomaly of the tricuspid valve, the origin of both vessels from the right ventricle, a tetralogy of Fallot with pulmonary atresia, tricuspid atresia, and occasionally from a truncus arteriosus or a single ventricle with reduced pulmonary blood flow

Operation for the relief of the pulmonary stenosis or to by pass the right ventricle lessens the incapacity of the patient but does not eliminate the basic abnormality

The prognosis is poor

Illustrative Cases

Case IX–1 G K (Harriet Lane Home, No A-66254) White female First seen at the Cardiac Clinic in October, 1948, at six years of age with the chief complaints of cyanosis and shortness of breath

History The cyanosis had been present since birth and was increasing in intensity A murmur and a thrill were first detected at one year of age The child was short of breath on mild exertion, but she did not squat when tired and she never had attacks of paroxysmal dyspnea

Physical examination The child was small and poorly developed although intensely cyanotic, she was bright and cooperative The heart was of approximately normal size, the heart sounds were of good quality At this time no murmurs were audible and the second sound at the base was not reduplicated

Laboratory data Red blood cell count 10 million hemoglobin 25 gm., hematocrit 85 per cent oxygen saturation of arterial blood 47 per cent

X-ray The heart was slightly enlarged with a concave curve at the base to the left of the sternum and reduced hilar markings (see Figure IX-3)

Electrocardiogram There was a right axis deviation, but there was also evidence of left as well as right ventricular hypertrophy in V_1 and the P waves were abnormally high (see Figure IX-6)

Clinical impression Although the absence of squatting and the absence of attacks of paroxysmal dyspnea were against the diagnosis of a tetralogy of Fallot, the child was obviously suffering from reduced pulmonary blood flow She was so sick that special studies were deemed dangerous

Subsequent course In October 1948 a subclavian pulmonary anastomosis was performed by Dr Alfred Blalock The child s color immediately improved but on the eighth postoperative day she developed right sided cardiac failure The heart became greatly enlarged The liver increased in size and pulsated The child developed pleural effusion ascites, and edema Digitalis and diuretics were of no avail repeated thoracenteses gave no relief

In January 1949 cardiac catheterization revealed the oxygen content of the blood in the superior vena cava to be 12 35 volumes per cent and that in the right auricle and right ventricle to be 13 0 and 13.27 volumes per cent respectively The pressure in the right ventricle was 110/27 mm of mercury The saturation of the arterial blood was 83 per cent. An angiocardiogram showed that the dye passed from the superior vena cava to the right auricle, where it lingered for a long time. The right ventricle was never well opacified but appeared to be small At the end of three seconds there was a suggestion of opacification of the main pulmonary artery and at the same time the aorta was faintly visualized The aorta appeared to arise from the left ventricle in its normal position

These studies indicated that the ventricular septum was intact and that the right ventricle was abnormally small In order to relieve the intractable right sided cardiac failure Dr Blalock created an auricular defect on January 29 A week later the patient developed auricular flutter with a rapid ventricular response Quinidine was tried but gave only temporary improvement The child lived for another month and then died of congestive cardiac failure

Autopsy No 21671 (performed by Dr Wharton) Interest was centered upon the heart The right auricle was tremendously dilated and hypertrophied Examination of the auricular septum showed that in addition to the large operative defect two of the margins of the foramen ovale were smoothly covered with endocardium thus indicating that prior to surgery the valve covering the foramen ovale had been patent The tricuspid valve was small and distorted by the very short papillary muscle (see Figure IX-1) The right ventricle was very small its wall was approximately the same thick

ness as that of the left ventricle The cavity of the right ventricle was small but not rudimentary The ventricular septum was intact There was a valvular pulmonary stenosis The cusps were fused together, forming a conical dome with a perforation which admitted only the tip of a small probe The pulmonary artery was small but ballooned out to a circumference of 2 6 cm The anastomosis was intact and its margins smoothly healed The pulmonary veins returned normally to the left auricle, the left auricle itself was normal except for the defect in its wall The mitral valve, the left ventricle, the aortic valves, and the aorta were all normal

Final anatomical diagnosis Pulmonary stenosis with an intact ventricular septum, hypertrophy of an underdeveloped right ventricle, dilatation and hypertrophy of the right auricle and patency of the foramen ovale, a surgical auricular defect, and anastomosis of the proximal end of the right subclavian artery to the side of the right pulmonary artery

Comment Although the basic malformation resembled that usually seen in pulmonic stenosis with an intact ventricular septum, the cavity of the right ventricle was much smaller In this instance the pulmonary stenosis was secondary to the defective development of the right ventricle The right ventricle was so small that, even if the pulmonary valve had been normal, the ventricle could not have pumped a normal volume of blood to the lungs

Defective development of the right ventricle may occur in combination with a complete transposition of the great vessels and a high ventricular septal defect, as illustrated in the following case report

CASE IX-2 Baby F (Harriet Lane Home, No A-23101) White female Admitted to the Harriet Lane Home in December 1941, at two days of age because of episodes of cyanosis

History Birth was reported as normal, with only an eight hour labor The baby was cyanotic at birth and there was great difficulty in resuscitation Respirations were established in fifteen minutes but the baby became cyanotic on nursing The following day she became intensely cyanotic and was brought to the hospital

Physical examination The infant s color was fairly good when she was quiet but became intensely cyanotic when she cried The heart rate was rapid, the sounds were of good quality There were no murmurs The lungs were clear No abnormalities were noted in the abdomen The extremities showed cyanosis but no edema

Laboratory data Red blood cell count 7 1 million white blood cell count 9,960 hemoglobin 28 gm The urine showed a trace of albumin and rare red blood cells and was loaded with epithelial cells and white blood cells The Wassermann reaction was negative



FIGURE IX-8 Defective development of the right ventricle combined with complete transposition of the great vessels and pulmonary atresia (same patient as in Figure IX-9) Infant

X ray The heart was boot shaped with an absence of the fullness of the pulmonary conus and a narrow shadow at the base to the left of the sternum (see Figure IX-8)

Electrocardiogram None was obtained

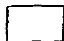
Clinical course The infant died at four days of age.

Clinical evaluation of the facts presented at a clinical pathological conference The contour of the heart was characteristic of a non functioning right ventricle. Nevertheless the early death of the patient pointed to a condition even more serious than that of a non functioning right ventricle with pulmonary atresia. Hence the author s diagnosis was a non functioning right ventricle combined with complete transposition of the great vessels and aortic atresia as such a malformation would cause great difficulty in the establishment of the systemic circulation

Autopsy No 1,695 (performed by Dr Sloan) The heart was small The superior vena cava and the inferior vena cava opened into the right auricle The tricuspid valve was small and opened into a diminutive right chamber The foramen ovale was covered by a valve but it was not sealed and had a tubular opening 0 5 cm in diameter, into the left auricle The pulmonary veins opened into the left auricle The mitral valve was normal and opened into a small but normally formed left ventricle There was a high ventricular septal defect The aorta arose from the right ventricle and par

Diagram IX–2

Head and upper extremities
Ductus Arteriosus
R Lung
S.V.C.
P.A.
Aorta
Pul. A
L Lung
PV
Pul V
L.A.
Remnant of Pul Art
R.A.
L.V.
R.V.
Descending Aorta
I.V.C.
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis

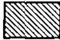

Venous blood

Diagram IX–2

Defective development of the right ventricle with the aorta arising mainly from the right ventricle pulmonary atresia an auricular septal defect a ventricular septal defect and a patent ductus arteriosus

In this malformation the development of the right ventricle is defective the aorta is transposed and slightly over rides the ventricular septum and the pulmonary artery, which arises from the left ventricle, is atretic at its base.

The blood from the right auricle flows in part into the right ventricle and in part through the auricular septal defect into the left auricle All the blood which enters the right ventricle is pumped out into the aorta Most of the blood in the aorta is directed to the systemic circulation and returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle. Inasmuch as there is pulmonary atresia the pulmonary pressure remains low, some blood from the aorta will flow through the ductus arteriosus to the lungs The blood which is directed to the lungs is returned by the pulmonary veins to the left auricle. Thus the left auricle receives a small amount of oxygenated blood from the lungs and also venous blood from the right auricle This admixture of oxygenated and venous blood is directed to the left ventricle The only way for the blood to be pumped out of the left ventricle is through the septal defect into the aorta which over rides the ventricular septum

In this malformation difficulty is encountered in the direction of blood to the lungs and also in the expulsion of blood from the left ventricle into the aorta

Clinical diagnosis The clinical findings are intense cyanosis and a characteristic contour of the heart The pulmonary atresia causes absence of the shadow normally cast by the pulmonary conus hence in the anterior posterior position, the shadow at the base of the heart to the left of the sternum is concave In addition in the left anterior-oblique position because of the defective development of the right ventricle, the margin of the cardiac shadow is straight

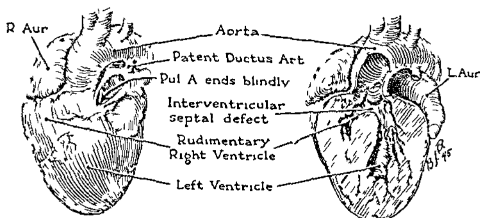


FIGURE IX–9 Defective development of the right ventricle combined with complete transposition of the great vessels and pulmonary atresia (same patient as in Figure IX–8) Infant

tially over rode the left ventricle so that blood from both ventricles was pumped out into the aorta The pulmonary artery was atretic at its base The ductus arteriosus was almost completely obliterated The course of the circulation is shown in Diagram IX–2 Figure IX–9 is a drawing of the gross specimen

Comment Although the anatomical structure was quite different from that antici pated, there was defective development of the right ventricle combined with a more serious condition than is usually found in association with a non functioning right ventricle The extreme dextroposition of the aorta rendered it difficult for the left ventricle to pump the blood out into the aorta The clarity of the lung fields should have suggested pulmonary atresia

B *Defect in the Musculature of the Right Ventricle Parchment Right Ventricle*

There is another anomaly of the right side of the heart in which the right ventricle exists as a relatively large chamber with an abnormally thin wall Two such cases have been reported in which the right ventricle was paper thin and contained virtually no heart muscle Uhl reported the case of a seven month old infant in whom autopsy showed virtually total absence of the myocardium of the right ventricle Castleman presented a similar case at one of the Massachusetts General Hospital clinical pathological conferences In Castleman s[3] case the woman lived for twenty four years and died of congestive failure with severe ascites

In this malformation, the right ventricle becomes a big, dilated, flabby cham

ber from which the blood is pumped slowly to the lungs As in all instances of abnormalities of the right side of the heart, the foramen ovale tends to be held open by the high pressure on the right side and thus a right to-left shunt is established at the auricular level

The author has seen two patients with maldevelopment of the wall of the right ventricle One was a boy of six who had shown cyanosis from birth and whose abnormality was incorrectly diagnosed as a tetralogy of Fallot Because of increasing incapacity he had had a Potts operation elsewhere He was referred to the author because of progressive cardiac enlargement and severe right sided cardiac failure The history revealed that as an infant he had never had paroxysmal dyspnea and that he did not squat when tired Autopsy revealed a large, dilated, paper thin right ventricle The myocardium of the right ventricle was virtually absent it was replaced by fat and fibrous tissue The thickened endocardium rested upon the epicardium Thus the right ventricle closely resembled the case reported by Uhl The left ventricle, however, showed some scarring and evidence of interstitial myocarditis

The other case, reported below, was carefully studied but as the patient is living, it cannot be certain that the two cases are identical *

Illustrative Case

CASE IX-3 H H (Harriet Lane Home, No B-49399) The infant was referred for diagnosis in March 1958 at the age of seventeen months

The family history was non-contributory with the only possible exception that the father was a genito urinary surgeon as was the father of one other patient with a defective development of the right ventricle †

History Growth and development were remarkably normal Except for slight persistent cyanosis the history was not significant until the day before admission, when the child had a mild convulsion and developed a left hemiplegia

Physical examination Temperature 38°C pulse 140 respirations 30, weight 8 3 kg height 73 cm The patient was well nourished and well developed There was slight persistent cyanosis which deepened on crying The left hemiplegia involved both the arm and the leg the eyes were deviated to the right The heart was enlarged The rhythm was regular There was no shock and no thrill The sounds were of fair quality and the only murmur audible was a low pitched mid-diastolic murmur at the

* See note on page 145

† Father s profession is mentioned only because of the extraordinary coincidence that the fathers of two patients with the same unusual malformation had the same occupation

apex, such as is commonly heard in a poorly functioning heart The liver was two finger breadths below the costal margin but did not pulsate The pulses were equal in all four extremities There was no edema

Laboratory data Hemoglobin 14 3 gm hematocrit 52 per cent

Teleoroentgenogram The heart was markedly enlarged and the lung fields were clear (see Figure IX–10)

Electrocardiogram The P waves in Lead II were abnormally high and there was a left axis deviation There was no evidence of right ventricular hypertrophy, even the normal dominance of the right ventricle in V_1 was absent (see Figure IX–11)

Initial clinical diagnosis Defective development of the right ventricle

Special tests Cardiac catheterization revealed that the oxygen saturation in the superior vena cava was 33 per cent, in the inferior vena cava 23 per cent, and in the right auricle varied between 25 and 30 per cent In the three samples obtained from the right ventricle the oxygen saturation was between 30 and 40 per cent, the pressure in that chamber was 16/2 mm of mercury The pulmonary artery was not entered The left auricle and the pulmonary veins were catheterized and the blood was found to be fully saturated The pressure in the left auricle was 12/6 mm of mercury and that in the right auricle was 10/3 mm of mercury The femoral arterial saturation was 57 per cent and the pressure was 110/57 mm of mercury

The low arterial oxygen saturation showed clear evidence of a right to-left shunt It also offered the probable explanation for the occurrence of the hemiplegia, inasmuch as cerebral thrombosis may occur in infants with either polycythemia or anoxemia The fact that the left auricle was catheterized showed that the foramen ovale was patent or that there was an auricular defect Although there was a slight increase in the oxygen saturation of the blood in the right ventricle as compared with that in the right auricle, the low pressure in the right ventricle clearly excluded a significant over riding of the aorta as the source of the right to left shunt For this reason it seemed probable that the entire right to left shunt was at the auricular level and that there was also a small left to right shunt at this level

Selective angiocardiography was performed in order to exclude conclusively a ven tricular septal defect First the catheter was placed in the outflow tract of the right ventricle and subsequently it was placed in the left auricle The right auricle was large, the tricuspid valve lay in its normal position The right ventricle was seen to be an enormously dilated extremely thin walled chamber (see Figure IX–12) The aorta was not visualized The great dilatation of the right ventricle beyond the tricuspid valve clearly differentiated this anomaly from Ebstein s anomaly of the tricuspid valve Fur thermore, there was an area at the apex of the heart into which dye never entered The left auricle appeared normal the left ventricle was displaced upward and occupied a horizontal position (see Figure IX–13) The left ventricle, however, showed evidence



Anterior posterior position



Left anterior-oblique position

FIGURE IX-10 Defective development of the musculature of the right ventricle (Case IX-3) Infant

apex, such as is commonly heard in a poorly functioning heart The liver was two finger breadths below the costal margin but did not pulsate The pulses were equal in all four extremities There was no edema

Laboratory data Hemoglobin 14 3 gm , hematocrit 52 per cent

Teleoroentgenogram The heart was markedly enlarged and the lung fields were clear (see Figure IX–10)

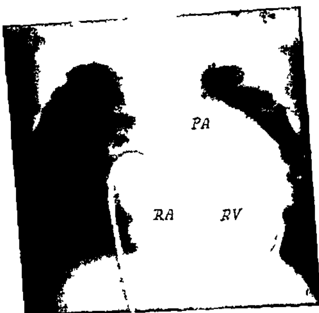
Electrocardiogram The P waves in Lead II were abnormally high and there was a left axis deviation There was no evidence of right ventricular hypertrophy, even the normal dominance of the right ventricle in V_1 was absent (see Figure IX–11)

Initial clinical diagnosis Defective development of the right ventricle

Special tests Cardiac catheterization revealed that the oxygen saturation in the superior vena cava was 33 per cent, in the inferior vena cava 23 per cent, and in the right auricle varied between 25 and 30 per cent In the three samples obtained from the right ventricle the oxygen saturation was between 30 and 40 per cent, the pressure in that chamber was 16/2 mm of mercury The pulmonary artery was not entered The left auricle and the pulmonary veins were catheterized and the blood was found to be fully saturated The pressure in the left auricle was 12/6 mm of mercury and that in the right auricle was 10/3 mm of mercury The femoral arterial saturation was 57 per cent and the pressure was 110/57 mm of mercury

The low arterial oxygen saturation showed clear evidence of a right to-left shunt It also offered the probable explanation for the occurrence of the hemiplegia, inasmuch as cerebral thrombosis may occur in infants with either polycythemia or anoxemia The fact that the left auricle was catheterized showed that the foramen ovale was patent or that there was an auricular defect Although there was a slight increase in the oxygen saturation of the blood in the right ventricle as compared with that in the right auricle the low pressure in the right ventricle clearly excluded a significant over riding of the aorta as the source of the right to left shunt For this reason it seemed probable that the entire right to left shunt was at the auricular level and that there was also a small left to right shunt at this level

Selective angiocardiography was performed in order to exclude conclusively a ven tricular septal defect First the catheter was placed in the outflow tract of the right ventricle and subsequently it was placed in the left auricle The right auricle was large the tricuspid valve lay in its normal position The right ventricle was seen to be an enormously dilated extremely thin walled chamber (see Figure IX–12) The aorta was not visualized The great dilatation of the right ventricle beyond the tricuspid valve clearly differentiated this anomaly from Ebstein s anomaly of the tricuspid valve Fur thermore, there was an area at the apex of the heart into which dye never entered The left auricle appeared normal, the left ventricle was displaced upward and occupied a horizontal position (see Figure IX–13) The left ventricle however, showed evidence



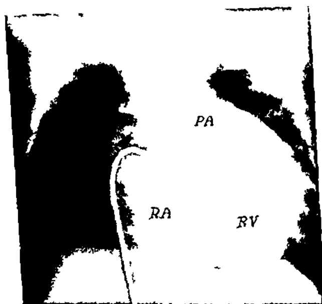


FIGURE IX-12 Defective development of the musculature of the right ventricle (Case IX-3) Infant

Dye is seen first in the pulmonary artery then in the right ventricle

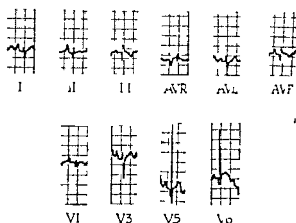


FIGURE IX–11 Defective development of the musculature of the right ventricle (Case IX–3) Infant

of normal ventricular contractions because there was a conspicuous difference in its size in systole and diastole

The angiocardiogram obviously did not show the exact nature of the abnormality The possibility of a tumor was considered, as it seemed well nigh impossible for a thin walled, greatly dilated chamber to displace the strong, thick walled left ventricle. A tumor in the septal wall might displace the left ventricle and either obstruct the circulation to the myocardium of the right ventricle or infiltrate its wall to such an extent as to destroy the musculature

Final clinical diagnosis Defective development of the wall of the right ventricle—a large thin walled chamber which was unable effectively to maintain the pulmonary circulation Left hemiplegia secondary to a cerebral thrombosis

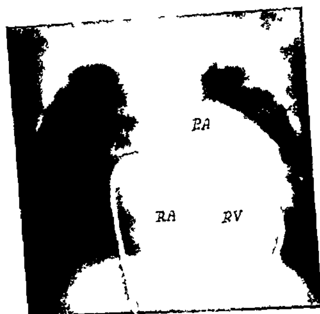
Treatment Glenn s[1] by pass operation was considered and so was vascularization of the right ventricle such as has been attempted for the left ventricle in infants in whom the left coronary artery arose anomalously from the pulmonary artery Inasmuch as either such operative procedure could be only palliative, operation was not recommended because the patient was in relatively good condition

Comment The exact nature of the malformation is not known It is however, clear that the pressure in both auricles was nearly the same as that in the right ventricle and that the right ventricle was a large chamber with such a low pressure that it was unable to pump the blood effectively to the lungs

The slight persistent cyanosis the high peaked P waves, and evidence of left ventricular preponderance in the electrocardiogram all indicated that the right ventricle, in spite of its size was seriously defective This was confirmed by the demonstration of a huge, thin walled chamber in which the pressure was low

Regardless of etiology, when the right ventricle is huge and its muscular wall is so thin that the electrocardiogram shows evidence of left ventricular dominance, the right ventricle is certainly grossly abnormal

This condition may be related to Sir William Osler s famous case of the



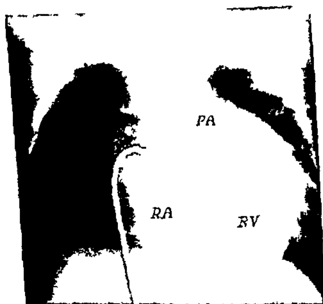


FIGURE IX-12 Defective development of the musculature of the right ventricle (Case IX-3) Infant

Dye is seen first in the pulmonary artery then in the right ventricle



Ventricular diastole



Ventricular systole

FIGURE IX–13 Defective development of the musculature of the right ventricle (Case IX–5) Infant

Dye is seen in the left side of the heart

parchment heart [4] Hence we have termed the condition a parchment right ventricle

References

1 Glenn W W L Circulatory by pass of the right side of the heart IV Shunt between superior vena cava and distal right pulmonary artery report of clinical application New England J Med 259 117–120, 1958
2 Uhl H S M Previously undescribed congenital malformation of heart almost total absence of the myocardium of right ventricle Bull Johns Hopkins Hosp 91 197–205 1952
3 Case 38201 *In* B Castleman and V W Towne (ed) Case Records of Massachusetts General Hospital New England J Med 246 785 1952
4 Segall H N Parchment heart (Osler) Am Heart J 40 948–950 1950

NOTE As this book was going to press the patient of Case IX–3 (see pages 139–144) had an intracardiac electrocardiogram which showed a tracing characteristic of Ebstein s anomaly of the tricuspid valve Subsequently a Glenn by pass operation was performed but the child did not survive

Autopsy showed extreme downward displacement of the tricuspid valve The wall of the right ventricle was paper thin and the foramen ovale was widely patent The structure of the heart was closely similar to that shown in Figure XIX–3 (see page 470) except that no blood could enter the apical portion of the right ventricle beneath the tricuspid valve The outflow tract of the right ventricle was a thin walled readily distensible chamber Indeed the entire wall of the right ventricle was thinner than that of the right auricle Even the septal wall appeared to be involved in the abnormality for at one point it was so thin as to be translucent

Comment The annulus at the tricuspid orifice had been mistaken for the tricuspid valve in the angiocardiogram The fullness of the pulmonary conus had also been considered to be inconsistent with Ebstein s anomaly The fullness was clearly due to the dilatation of the outflow tract.

The involvement of the entire right ventricle musculature suggests that Ebstein s anomaly may be related to the parchment right ventricle except that in the latter the tricuspid valve is normal Clearly the two conditions call for differentiation In this instance the intracardiac electrocardiogram gave the clue to the diagnosis but at the time of the report we have no knowledge of the intracardiac electrocardiographic pattern of a parchment right ventricle

CHAPTER X

COMPLETE TRANSPOSITION OF THE GREAT VESSELS

COMPLETE transposition of the great vessels means that the aorta arises from the right ventricle and the pulmonary artery from the left ventricle This may occur as an isolated anomaly It is, however, frequently associated with an auricular or a ventricular septal defect and may be associated with almost any cardiac abnormality, as for example, a single ventricle In the latter condition the great vessels arise from a common chamber but the aorta lies anterior to the pulmonary artery

Furthermore, the pulmonary artery may be of normal size, it may be greatly dilated, or it may be stenotic or even atretic Although the same is theoretically true as regards the aorta, it is usual for the aorta to be of normal size unless the anomaly is associated with some gross malformation which dominates the clinical syndrome, such as aortic atresia

The condition is more frequently compatible with life when the pulmonary artery is greatly dilated or markedly stenosed than when both great vessels are of equal size Nevertheless, the basic principles and the underlying hemodynamics are the same in most types of transposition of the great vessels Therefore let us first consider a complete transposition of the great vessels in which both great vessels are of normal size, and then the factors which aid in the differentiation of the other types

ETIOLOGY

The etiology of all types is obscure Much evidence has been brought forward to show that the condition results from an abnormal torsion of the aortic septum whereby the aortic septum fails to meet the ventricular septum in the normal manner For a discussion of the etiology of this condition, the reader is referred to the works of Rokitansky,[1] Spitzer,[2, 3] Pernkopf and Wirtinger,[4] Harris and Farber,[5] Bremer,[6] Emerson and Green,[7] and Lev and Saphir [8]

A *Complete Transposition of the Great Vessels with Both Great Vessels of Approximately Normal Size*

NATURE OF THE MALFORMATION

The essential feature of this malformation is that the aorta arises from the right ventricle and the pulmonary artery from the left ventricle The coronary

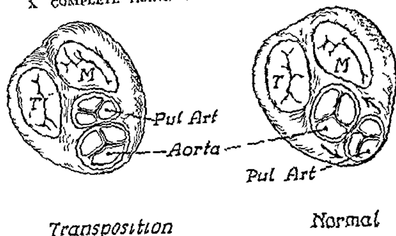


FIGURE X-1 Complete *transposition* of the great vessels and normal heart
The drawing shows the base of the heart with the auricles removed

arteries arise from the base of the *aorta*, which is usually of normal size The aorta is *rotated forward and to the left* and the pulmonary artery is rotated backward and to the right in a counterclockwise direction Only *rarely is the aorta* rotated so far to the right that it occupies the position of the normal pulmonary artery Usually, although the aorta arises entirely from the right ventricle, it lies above the *mid portion* of that chamber and in most instances the pulmonary artery comes to lie directly posterior to the aorta (*see Figure X-1*)

Some defect within the heart, either a ventricular septal defect or an auricular septal defect, is *generally present* Indeed, in order *for this malformation* to be compatible with life, even for a short time, *some pathway* must exist which will permit at least a slight interchange of blood between the systemic and pulmonary circulations If neither *the ductus* arteriosus nor the *foramen* ovale is patent, there must *be an auricular* or a ventricular *septal defect*, or an aortic septal *defect, or an anomaly* of the venous *return*, or else some combination of these anomalies

At birth the ductus arteriosus and the foramen ovale *are both* normally patent In cases of complete transposition of the *great vessels in* which there *is no* other abnormality, these pathways continue after birth to be of functional importance and to play a vital role in the anomalous circulation *Indeed, the* question of prime importance to *extra uterine* life is *whether there is* the possibility of *some* admixture of venous and arterial blood *within the* auricles or within the ventricles, or whether *life is* dependent upon the patency of the ductus arteriosus and the *foramen ovale*

The structure of the heart in a *complete transposition* of the great vessels

with an intact ventricular septum is shown in Figure x–2 In this instance the only additional malformation which permitted the shunting of blood from one circulation to the other was a small aortic septal defect

The drawing of a complete transposition of the great vessels with a ventricular septal defect is shown in Figure x–3

COURSE OF THE CIRCULATION

During fetal life, when the great vessels are transposed, the right ventricle pumps the blood to the head and the upper extremities and some of the blood to the trunk and to the lower extremities and the left ventricle meets the high pressure of the unexpanded lungs and sends blood through the ductus arteriosus to the trunk and lower extremities Consequently the altered position of the great vessels causes no significant change in the course of the fetal circulation or in the amount of work required of the two ventricles (see Figure x–4) At birth the heart is normal in size Indeed, the condition is so inconsequential to fetal life that the baby ordinarily lives till term For this reason a complete transposition of the great vessels is a relatively common malformation

After birth the situation is quite different With the expansion of the lungs and the establishment of respiration, the left ventricle pumps the blood through the pulmonary artery to the lungs, whence it is returned in the normal fashion by the pulmonary veins to the left auricle The blood from the right ventricle is pumped out into the aorta and is returned by the superior vena cava and the inferior vena cava to the right auricle Thus there is a tendency for the blood to be pumped around and around the systemic circulation and in a similar manner around the pulmonary circulation

If there are no intracardiac abnormalities, the foramen ovale and the ductus arteriosus, which are normally patent at birth, are the only possible pathways for the crossing of the two circulations As is always the case, with the first breaths of life, blood will flow from the aorta to the pulmonary artery, the blood which is directed to the lungs is returned to the left auricle and the left ventricle Inasmuch as the valve which covers the foramen ovale closes from left to right, all the blood from the left auricle will flow into the left ventricle and from there it is again pumped to the lungs Thus the blood re circulates through the lesser circulation As long as the pulmonary pressure remains low, some blood from the aorta flows through the ductus arteriosus to the lungs Since all the blood which goes to the lungs is returned to the left side of the heart the pressure in the left ventricle and in the pulmonary artery will eventually rise

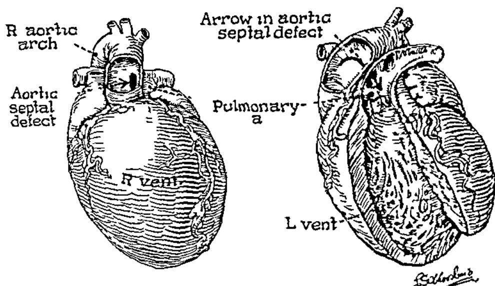


FIGURE X-2 Complete transposition of the great vessels with an intact ventricular septum and an aortic septal defect

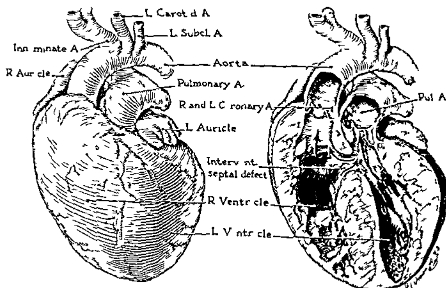


FIGURE X-3 Complete transposition of the great vessels with a high ventricular septal defect

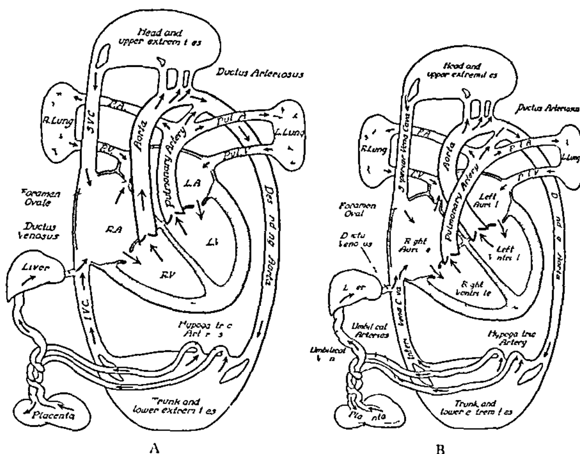


FIGURE X-4 Fetal circulation (A) Complete transposition of the great vessels with a patent ductus arteriosus and a patent foramen ovale and (B) normal heart

When the pressure in the pulmonary artery exceeds that in the aorta, the direction of the flow of blood through the ductus arteriosus will be reversed and blood will flow from the pulmonary artery to the systemic circulation The blood so shunted is returned to the right auricle Thereupon the pressure on the right side of the heart will rise, while that on the left will fall As soon as the pressure in the right auricle exceeds that in the left auricle the valve covering the foramen ovale will be forced open and blood will flow from the right auricle to the left, thereby again raising the pressure in the left auricle and the left ventricle and the pulmonary artery Thereafter blood will continue to flow from the pulmonary artery through the ductus arteriosus to the descending aorta and from the right auricle through the foramen ovale and the left auricle This mechanism, which is shown in Diagram x-1, permits the continuous crossing of the two circulations so long as the ductus arteriosus and the foramen ovale remain patent As these two pathways undergo normal obliteration, the condition becomes

incompatible with life Infants with this condition usually die within the first month

For an infant with a complete transposition of the great vessels *to survive the closure of fetal pathways*, there must be an additional defect, either an auricular or a ventricular septal defect or both (see Diagram x–2) or even, in rare instances, an aortic septal defect The transposition of the great vessels causes the blood which is pumped out from the right ventricle into the aorta to be returned by the superior vena cava and the inferior vena cava to the right auricle, and that which is pumped out from the left ventricle into the pulmonary artery to be returned by the pulmonary veins to the left auricle Consequently, even though a normal volume of blood reaches the lungs, difficulty is encountered in the direction of venous blood to the lungs and of oxygenated blood to the systemic circulation Hence, in this malformation, the greater the volume of the shunt, the greater is the admixture of arterial and venous blood and the less intense is the cyanosis

Furthermore, if blood is shunted from the right side of the heart to the left, it circulates through the lungs and is returned to the left side of the heart, whereas, if it is shunted from the left side to the right, it circulates through the body and is returned to the right side of the heart In other words, the blood which is shunted from one side to the other is returned to the side to which it was shunted (see Figure x–5) Consequently the blood tends to pile up on one side of the circulation There is, however, a limit to the amount of the shunt in one direction, because while the pressure on one side steadily rises, that on the other side must fall Whenever the pressure on one side exceeds that on the other the direction of the shunt will be reversed

When the blood is shunted from the lungs to the systemic circulation, it is easy to understand that the pressure in the systemic circulation will soon exceed that in the pulmonary circulation and that the direction of the shunt will be reversed Thereupon blood will be shunted from the systemic circulation to the lungs until the pressure in the pulmonary circulation exceeds that in the systemic circulation As soon as the pressure in the pulmonary circulation exceeds that in the systemic circulation blood will again be shunted to the systemic circulation When there is but a single defect, the reversal in the direction of the shunt is the only mechanism which permits the return of blood to the side *from* which it was shunted Moreover, the direction of oxygenated blood to the systemic circulation and of venous blood to the lungs is dependent upon the perpetual reversal in the direction of the shunt This mechanism of the piling up

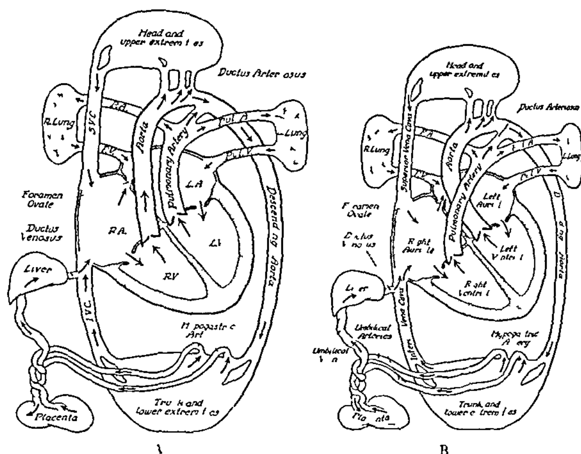


FIGURE X-4 Fetal circulation (A) Complete transposition of the great vessels with a patent ductus arteriosus and a patent foramen ovale and (B) normal heart

When the pressure in the pulmonary artery exceeds that in the aorta, the direction of the flow of blood through the ductus arteriosus will be reversed and blood will flow from the pulmonary artery to the systemic circulation The blood so shunted is returned to the right auricle Thereupon the pressure on the right side of the heart will rise, while that on the left will fall As soon as the pressure in the right auricle exceeds that in the left auricle the valve covering the foramen ovale will be forced open and blood will flow from the right auricle to the left, thereby again raising the pressure in the left auricle and the left ventricle and the pulmonary artery Thereafter blood will continue to flow from the pulmonary artery through the ductus arteriosus to the descending aorta and from the right auricle through the foramen ovale and the left auricle This mechanism, which is shown in Diagram X-1, permits the continuous crossing of the two circulations so long as the ductus arteriosus and the foramen ovale remain patent As these two pathways undergo normal obliteration, the condition becomes

DIAGRAM X-1

Complete transposition of the great vessels with a patent ductus arteriosus and a patent foramen ovale

In this malformation there is complete transposition of the great vessels The ductus arteriosus is patent and the foramen ovale is covered by a valve which is held open by the high pressure in the right auricle and permits the blood to flow in one direction only namely, from right to left

The blood from the right auricle flows into the right ventricle, is pumped out through the aorta to the systemic circulation, and is returned by the superior and inferior venae cavae to the right auricle The blood from the left auricle flows into the left ventricle and is pumped out through the pulmonary artery to the lungs whence it is returned by the pulmonary veins to the left auricle

Inasmuch as the valve covering the foramen ovale closes from left to right, blood can be shunted only through the foramen ovale from right to left The blood so shunted raises the pressure in the left auricle and the left ventricle and in the pulmonary artery Therefore some blood from the pulmonary artery flows through the ductus arteriosus to the descending aorta The blood so shunted is returned to the right auricle thus raising the pressure in the right auricle. The foramen ovale is again forced open and some blood from the right auricle flows to the left auricle and thence to the left ventricle There the cycle starts again

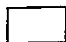
Clinical diagnosis The heart becomes greatly enlarged In the anterior posterior position there is an absence of the shadow cast by the *pulmonary conus* and the shadow cast by the great vessels is narrow, this shadow increases in width when viewed in the left anterior-oblique position Murmurs are of no diagnostic aid The electrocardiogram usually shows a right axis deviation and evidence of *right ventricular hypertrophy*

Cyanosis is intense Inasmuch as the direction *of the flow of blood* is *from the* pulmonary artery through the ductus arteriosus to the *lower extremities* the left ventricle is pumping against systemic pressure consequently there is *pulmonary* hypertension Furthermore some arterial blood is directed through the ductus arteriosus to the descending aorta hence the *lower extremities* are less cyanotic than the upper extremities The line of demarcation of the *cyanosis* lies at the brim of the pelvis

The double lines as shown in the lower *right hand* block indicate that the cyanosis is of greater intensity in the head and the *upper* extremities than in the lower extremities

DIAGRAM X–1

Head and upper extremities
Ductus Arteriosus
R' Lung
S.V.C.
P.A.
Pul. A
L Lung
PV
Pul V
Aorta
Pulmonary Artery
L.A.
Descending
Foramen Ovale
R.A.
L.V.
R.V.
I.V.C.
Aorta
Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood with but slight admixture of oxygenated blood



Venous blood

DIAGRAM X-2

Complete transposition of the great vessels with auricular and ventricular septal defects and a patent ductus arteriosus

n this malformation the aorta arises from the right ventricle and the pulmonary ar ery from the left ventricle In addition, there is a gross defect in both the auricular eptum and the ventricular septum The ductus arteriosus may remain patent

The blood from the right auricle flows into the right ventricle and can be pumped out into either the aorta or the pulmonary artery Inasmuch as the aorta arises from the right ventricle the greater part of the blood which enters the right ventricle is pumped out through the aorta to the systemic circulation All the blood in the systemic circulation is returned by way of the superior and inferior venae cavae to the right auricle Similarly the blood which passes from the left auricle to the left ventricle can leave by way of the pulmonary artery or by way of the aorta Inasmuch as the pulmonary artery arises directly from the left ventricle, most of the blood in the left ventricle is pumped into the pulmonary artery This blood goes to the lungs for oxygenation and the oxygenated blood is returned to the left auricle and thence to the left ventricle

The high ventricular septal defect offers easy opportunity for some oxygenated blood from the left side of the heart to be pumped out into the aorta The blood so shunted is returned to the right auricle and the right ventricle As the pressure on the right side rises blood will be shunted from right to left through both the auricular septal defect and the ventricular septal defect Thereby venous blood is directed to the lungs for oxygenation As in all cases of complete transposition of the great vessels, there is always pulmonary hypertension In this malformation there is ample opportunity for shunting blood from one side of the heart to the other and equal opportunity for a reversal in the direction of the shunt The direction of the flow of blood through the ductus arteriosus will depend upon the relative pressures in the aorta and the pulmonary artery However the crossing of the two circulations is not dependent upon this pathway Therefore the condition is compatible with life after the closure of the ductus arteriosus the ductus arteriosus may or may not be patent

Clinical diagnosis is based upon the size and the contour of the heart The heart is enlarged both ventricles are huge There is absence of fullness of the pulmonary conus and the shadow cast by the great vessels is narrow This shadow increases in width when viewed in the left anterior-oblique position There may be evidence of congestion in the lungs Murmurs are of no aid in diagnosis The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

Diagram X-2

Head and upper extremities
Ductus Arteriosus
R Lung
S.V.C.
P.A.
P.V.
Aorta
Pulmonary Art.
Pul. A.
L Lung
Pul V
L.A.
R.A.
L.V.
R.V.
Descending Aorta
I.V.C.
Trunk and lower extremities



Arterial bl[illegible] (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

is pulmonary stenosis or great dilatation of the pulmonary artery than when the pulmonary artery is of normal size If such a balance is established, although the heart is enlarged, the enlargement is not progressive and the condition may be compatible with life for a number of years (see Sections B and C)

PHYSIOLOGY OF THE MALFORMATION

In this malformation, instead of the two circulations crossing in the normal way, the blood is directed around and around the systemic circulation and similarly around the pulmonary circulation Moreover, it is venous blood which circulates through the systemic circulation and oxygenated blood which circulates through the lungs Consequently, although some crossing of the two circulations is essential for life, there is real difficulty in the direction of venous blood to the lungs The effective flow is very small Furthermore, in this malformation the greater the volume of the shunt, the greater is the volume of venous blood directed to the lungs and the greater is the volume of arterial blood which can be directed to the body Hence the greater the shunt, the less intense is the cyanosis

As previously mentioned, the reversal in the direction of the shunt occurs when the pressure in one circulation exceeds that of the other, hence a balance is established when the pressure in the pulmonary circulation approximates that in the systemic circulation In other words, severe pulmonary hypertension is the rule This finding has been repeatedly confirmed at operation Indeed, the author had thought that an end to-end systemic pulmonary anastomosis, in which the pulmonary artery is severed, would reduce the pressure in the distal segment sufficiently so that the anastomosis might aid in the direction of blood to the lungs Dr Alfred Blalock performed several such operations It was, however, found that the back pressure in the lungs was so high that the anastomosis between the proximal end of the subclavian artery and the distal end of the pulmonary artery could not function Indeed, a systemic pulmonary anastomosis is of no avail except when there is pulmonary stenosis and the pressure in the pulmonary artery is low

Edwards[9] believes and the author agrees, that in complete transposition of the great vessels, as in malformations where both great vessels arise from the same ventricle, the resistance in the pulmonary vascular bed probably regulates the amount of blood which is directed to the two circulations Be that as it may, in complete transposition of the great vessels, except when there is also pulmonary stenosis, pulmonary hypertension is the rule

It is worthy of note that, although the pulmonary blood flow may be exces-

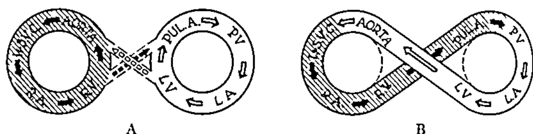


Figure X-5 (A) Course of the circulation in a complete transposition of the great vessels and (B) normal crossing of the two circulations

of blood first on one side and then on the other places an ever increasing strain on both sides of the heart, both ventricles undergo progressive dilatation and hypertrophy

The course of the circulation with a ventricular septal defect is shown in Diagram X-3 Defects in the auricular septum are usually larger than those in the ventricular septum Moreover, inasmuch as the pressure in the auricles is far lower than that in the ventricles and the difference between the pressures in the two auricles is slight, a slight increase in the pressure in either auricle represents a far greater percentage of change than a similar rise in pressure in the ventricles Therefore the crossing of the two circulations occurs more easily with an auricular septal defect than with a ventricular septal defect (see Diagram X-4) For this reason infants with a complete transposition of the great vessels usually do better when the condition is associated with a gross defect in the auricular septum than when it is associated with a ventricular septal defect

There are a few individuals with complete transposition of the great vessels in whom the defects within the heart are such that a balance is established which enables the patient to live for a number of years For this to be possible the load placed on the heart must be constant In the author s opinion such a balance is usually established by a figure of eight shunt in which the volume of the shunt from left to right is of the same magnitude as that from right to left Such a circulation can more readily be established when there are two associated anomalies than when only one is present When there is but a single defect, the crossing between the two circulations must occur through that defect and a perpetual reversal in the direction of the shunt is common When there are two defects, the shunt from left to right may be through one defect and that from right to left through the other, thereby permitting some degree of crossing of the two circulations When the two shunts are of equal volume, the load placed on the heart is constant Such a mechanism appears to be more easily established when there

DIAGRAM X-3

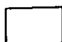
Complete transposition of the great vessels with a high ventricular septal defect

In this malformation the aorta arises from the right ventricle, the pulmonary artery from the left ventricle. There is in addition a high ventricular septal defect

The blood *from the right* auricle flows into the right ventricle, from there most of the blood is pumped out through the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle Part of the blood from the right ventricle is pumped through the septal defect into the pulmonary artery Thus venous blood from the right ventricle is mixed with the oxygenated blood from the left ventricle and flows to the lungs, where the blood is oxygenated It is, however, only the venous blood from the right ventricle which is able to take up oxygen All the blood from the lungs is returned by the pulmonary veins to the left auricle, thence it flows to the left ventricle and is again pumped out into the pulmonary artery Just as some blood from the right ventricle may pass through the septal defect to the pulmonary artery so some blood from the left ventricle may pass through the defect into the aorta In whichever direction the blood is shunted it is returned to that same side This mechanism tends to increase the pressure on the side to which the blood is shunted and lower the pressure on the side from which the blood is shunted Hence the relative pressure on the two sides of the heart is reversed Consequently the direction of the shunt is reversed Thus each side of the heart alternately has an excessive amount of work Both ventricles undergo progressive enlargement It is the relative pressure in the two circulations which regulates the direction and the volume of the shunt The systemic and pulmonary pressures are approximately equal, there is marked pulmonary hypertension Great difficulty however, is encountered in the direction of the oxygenated blood to the systemic circulation

Clinical diagnosis Cyanosis is intense Clubbing develops at an early age The heart undergoes progressive enlargement A systolic murmur and a gallop rhythm are the rule Cardiac failure occurs early with congestion in the lungs engorgement of the liver and edema of the extremities The contour of the heart is egg shaped with a narrow pedicle at the base which increases in width in the left anterior-oblique position The vascular markings extend nearly to the periphery of the lungs The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

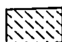
Diagram x–3



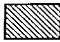
Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

DIAGRAM X-4

Complete transposition of the great vessels with an auricular septal defect

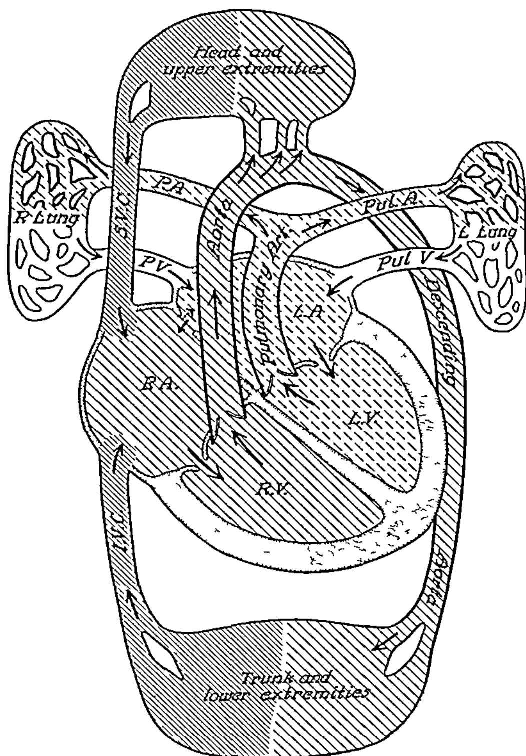
In this malformation the aorta arises from the right ventricle, the pulmonary artery from the left ventricle and there is a gross defect in the auricular septum The ductus arteriosus undergoes normal obliteration

The blood from the right auricle flows into the right ventricle and is pumped out by way of the aorta to the systemic circulation, thence it is returned by the superior and inferior venae cavae to the right auricle. The blood from the left auricle flows into the left ventricle and is pumped out through the pulmonary artery to the lungs whence it is returned by the pulmonary veins to the left auricle The gross defect in the auricular septum permits the flow of blood in either direction consequently, as the lungs expand and blood flows readily from the left ventricle to the lungs and also through the ductus arteriosus to the lungs the volume of blood which flows to the lungs is increased This increased volume of blood is returned to the left auricle. Therefore the pressure in that chamber will rise until it exceeds that of the right auricle, whereupon the direction of the shunt will be reversed There the cycle starts again Because of the low pressure in the auricles a very slight increase or decrease in the pressure in either auricle may alter the direction and the volume of the shunt Such changes occur regardless of whether the ductus arteriosus is patent Nevertheless an equilibrium cannot be established until the pulmonary pressure is approximately equal to the systemic pressure Consequently there must be pulmonary hypertension

Clinical diagnosis Cyanosis is obvious but may not be as intense as it is in other types of transposition of the great vessels The heart is enlarged There is absence of fullness of the pulmonary conus In the anterior posterior position the shadow cast by the great vessels is narrow this shadow increases in width when viewed in the left anterior-oblique position Murmurs are of no diagnostic significance The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

If the defect in the auricular septum is relatively large, the low pressure in the auricle permits the ready shunting of blood Inasmuch as any increase in the shunt in either direction is beneficial, a balance may be established with a relatively good pulmonary blood flow and proportionally less cyanosis Under such circumstances the condition may be compatible with life for a number of years

Diagram X-4



Legend		Legend	
(blank box)	Arterial blood (fully saturated)	(hatched box)	Venous and arterial blood Cyanosis visible
(dashed hatched box)	Small admixture of venous blood No visible cyanosis	(dense hatched box)	Venous blood

arteries, both of which arise from the subclavian arteries, the line of demarcation of the cyanosis lies at the brim of the pelvis

This distribution of cyanosis *is most conspicuous* when a complete transposition of the great vessels is combined with *complete interruption* of the isthmus of the aorta (see Section D and Diagram X-9) *It is, however, most frequently* seen in young infants with a complete transposition of the great vessels in whom the intracardiac defect is small or absent and both the ductus arteriosus and the foramen ovale are patent

Clubbing of the extremities develops as the red blood cell count rises It is usually apparent by one year of age

Polycythemia steadily increases The red blood cell count may approach 10 million and there is a proportionate increase in the amount of available hemoglobin and in the hematocrit reading If the duration of life permits, the secondary changes in the blood usual with long standing polycythemia develop the blood platelets and the blood fibrinogen are decreased and the blood clot becomes friable

Respirations are rapid and shallow

Barrel shaped chest deformity develops, owing to air hunger and the excessive rapidity of the respirations

Repeated episodes of loss of consciousness may occur but attacks of paroxysmal dyspnea are rare The pressures in the two circulations are approximately equal and there is no abrupt change in the flow of blood to the lungs *The blood* gradually piles up first on one side and then on the other The infant suffers from progressive anoxemia until he finally loses consciousness Indeed, he may die from anoxemia unless the shunt reverses, whereupon he gradually improves

Difficulty in feeding and failure to gain are common complaints Weight gain is usually extremely slow because the infant can digest only a small amount of food at a time and simply cannot eat enough to meet the normal requirements of the body

Growth and development are retarded The baby is slow to hold up his head alone, to turn over, and to crawl Few of these children learn to walk before three or four years of age Their exercise tolerance is extremely limited and they quat when tired Their growth may be extremely stunted (see Section C)

Pulmonary congestion is of common occurrence unless there is an associated pulmonary stenosis Usually plenty of blood reaches the lungs Consequently, as compensation fails, the patient usually develops râles in the lungs

The liver becomes engorged, it not infrequently descends to the umbilicus

sive, over a period of years collateral circulation does develop Inasmuch as the aorta carries venous blood, such collateral circulation aids in the direction of venous blood to the lungs

CLINICAL FINDINGS

The outstanding clinical manifestations are due to anoxemia, polycythemia, and difficulty in the direction of oxygenated blood to the systemic circulation

Cyanosis may or may not be apparent at birth Regardless of the structure of the heart, the first breaths of life direct the blood to the lungs If the cardiac septa are intact, great difficulty is encountered in the direction of the oxygenated blood to the systemic circulation Cyanosis appears early and is intense If, however, there is free communication between the two sides of the heart, that is, if there is a gross defect in either the ventricular or the auricular septum, infants with this malformation may seem remarkably normal during the neonatal period Cyanosis may be entirely absent, or it may be apparent only when the baby cries or nurses Indeed, such a baby may be discharged from the nursery as a normal infant with a normal weight gain Nevertheless, inasmuch as the right ventricle pumps the blood around and around the systemic circulation, sooner or later cyanosis almost invariably develops and becomes progressively more intense

The author has, however, seen two patients with complete transposition of the great vessels who showed no cyanosis even in childhood Both were operated on because of large ventricular septal defects In one instance there was a gross defect in the auricular septum and in the other the ventricular septum was deviated so that, although the great vessels occupied their normal positions, the pulmonary artery arose from the left ventricle and the aorta arose from the right ventricle (for the findings upon cardiac catheterization, see below under Special Tests)

The distribution of the cyanosis is often of great diagnostic significance In this malformation the blood from the left ventricle may be pumped from the pulmonary artery through the ductus arteriosus to the descending aorta Under such circumstances oxygenated blood from the left ventricle is directed to the trunk and the lower extremities (see Diagram X-1) Consequently the lower extremities will be less cyanotic than the upper The difference in cyanosis, though definite, is not conspicuous and is best appreciated by placing the infant's hand beside his foot Inasmuch as the skin of the thorax and abdomen receives its nutrition through the internal mammary arteries and the superficial epigastric



At one month



At eight and one half months

FIGURE X-6 Complete transposition of the great vessels with a high ventricular septal defect and a situs inversus Anterior posterior position shown in reverse

The reversal of the x rays makes it easier to compare the contours with those of the same malformation in a heart that occupies its normal position

and may extend to the brim of the pelvis There is seldom either tricuspid insufficiency or stenosis, hence the liver does not pulsate

Edema of the extremities occurs with cardiac failure

The pulse pressure is narrow, *the blood pressure* is often difficult to obtain

CARDIAC FINDINGS

The heart at birth is normal in size Unless the duration of life is too short to permit the heart to enlarge, the malformation usually leads to progressive cardiac enlargement The greater the difficulty in the crossing of the two circulations, the sooner the signs of distress become apparent and the more rapidly the heart enlarges Inasmuch as the right ventricle pumps the blood throughout the systemic circulation, that ventricle becomes huge The left ventricle also carries a full load It pumps the blood around and around the pulmonary circulation under abnormally high pressure, in addition, while the ductus arteriosus remains patent, the left ventricle also pumps some blood through the ductus arteriosus to the descending aorta Hence the left ventricle also becomes greatly enlarged The rate at which the heart enlarges varies with the associated anomalies In most instances, however, the heart becomes enormously enlarged between seven and ten months of age, as illustrated in Figures X-6 and 7 Figures X-8 and 9 show the oblique views of the same infant

Nevertheless, occasionally a balance is established, cardiac enlargement comes to an end Although theoretically this may occur after any degree of enlargement, the patient is far more likely to survive if a balance is established promptly Consequently in patients who live to childhood and early adult life the heart is usually only slightly enlarged

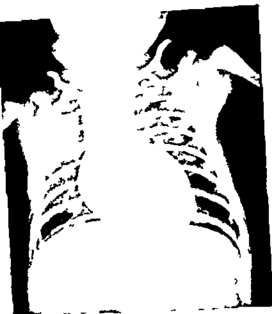
The heart sounds are forceful The *second sound* over the pulmonary area is usually accentuated, as the aorta lies far to the left

Murmurs and thrills are of no diagnostic importance The quality and intensity of both the murmur and the thrill vary with the nature of the concomitant malformation and also with the relative pressure in the two circulations Usually the systolic murmur is not very intense Inasmuch as the pressures in the two circulations are approximately equal, the systolic murmur generally lacks the rasping quality characteristic of a ventricular septal defect For the same reason, even if the ductus arteriosus is patent, a continuous murmur never develops

A thrill may or may not be palpable over the precordium

A gallop rhythm is frequently audible, as compensation is usually precarious

Cardiac failure is common In most instances, the heart undergoes progres



At one month



At eight and one half months

FIGURE X-6 Complete transposition of the great vessels with a high ventricular septal defect and a situs inversus Anterior posterior position shown in reverse

The reversal of the x rays makes it easier to compare the contours with those of the same malformation in a heart that occupies its normal position

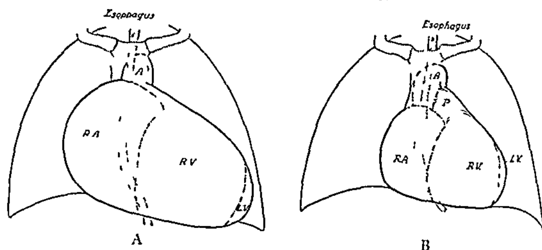


FIGURE X-7 (A) Complete transposition of the great vessels with a high ventricular septal defect and (B) normal heart Infant

sive enlargement and eventually becomes enormous The liver is enlarged and, inasmuch as the circulation to the lungs is adequate, pulmonary congestion and rales in the lungs are common Indeed, this is the outstanding malformation of early infancy in which one finds persistent cyanosis and cardiac failure with engorgement of the liver, edema of the extremities, and congestion of the lungs These infants usually die before eighteen months of age Death is due either to anoxia or cardiac failure combined with pulmonary congestion

X RAY AND FLUOROSCOPIC FINDINGS

The contour of the heart varies with the age of the patient and the size of the pulmonary artery Usually all four chambers of the heart are greatly enlarged The enlargement extends both to the right and to the left

In infancy the contour of the heart frequently resembles an egg lying on its side with the tip pointing slightly downward and to the left, the base of the egg, which lies to the right of the sternum, is formed by the dilatation of the right auricle (see Figure X-6, also Figures X-10 and 11) Although the aorta arises from the right ventricle, it seldom arises as far to the left as does the normal pulmonary artery Hence in the anterior posterior position it is usual to find a slight concavity rather than a convexity of the upper border of the cardiac silhouette to the left of the sternum

In the left anterior oblique position both ventricles are seen to be enlarged In an estimation of the size of the right ventricle, it is important to remember that in this malformation the aorta is displaced anteriorly For this reason the increase in the size of the right ventricle relative to the aorta is less pronounced



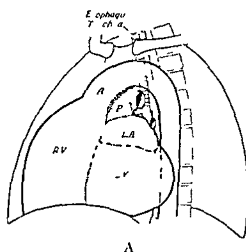
Right anterior-oblique position shown in reverse



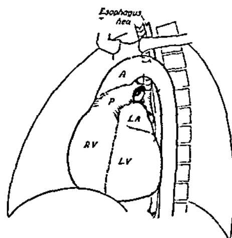
Left anterior-oblique position shown in reverse

Figure x-8 Complete *transposition* of the great vessels with a high ventricular septal defect and a situs inversus (same patient as Figure x-6) At eight and one half months

The reversal of the x rays makes it easier to compare the contours with those of the same malformation in a heart that occupies its normal position

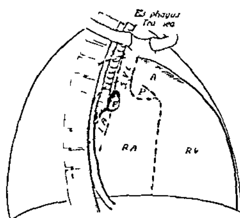


A

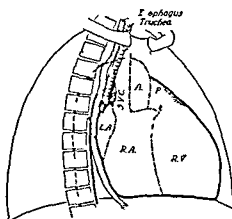


B

LEFT ANTERIOR-OBLIQUE POSITION



A



B

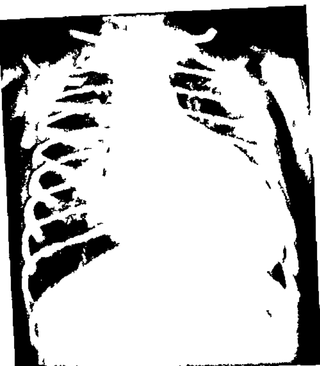
RIGHT ANTERIOR OBLIQUE POSITION

FIGURE X-9 (A) Complete transposition of the great vessels with a high ventricular septal defect and (B) normal heart Infant

than in other malformations Both the aorta and the right ventricle, however, extend closer to the anterior chest wall than in the normal heart (see Figures X-8 through 11) Terminally it is common to find that the right ventricle nearly touches the anterior chest wall and that, when the patient is rotated to an angle of 60°, the left ventricle extends posterior to the spinal column

In the right anterior oblique position the heart frequently appears to fill the entire chest The entire esophagus may be displaced backward but there is no specific enlargement of the left auricle (see Figures X-8 and 9)

The altered positions of the great vessels relative to the ventricles cause a characteristic change in the shadow cast by these vessels [10] When the aorta arises



Anterior posterior position



Left anterior-oblique position

FIGURE X-10 Complete transposition of the great vessels with a patent ductus arteriosus and a patent foramen ovale Infant

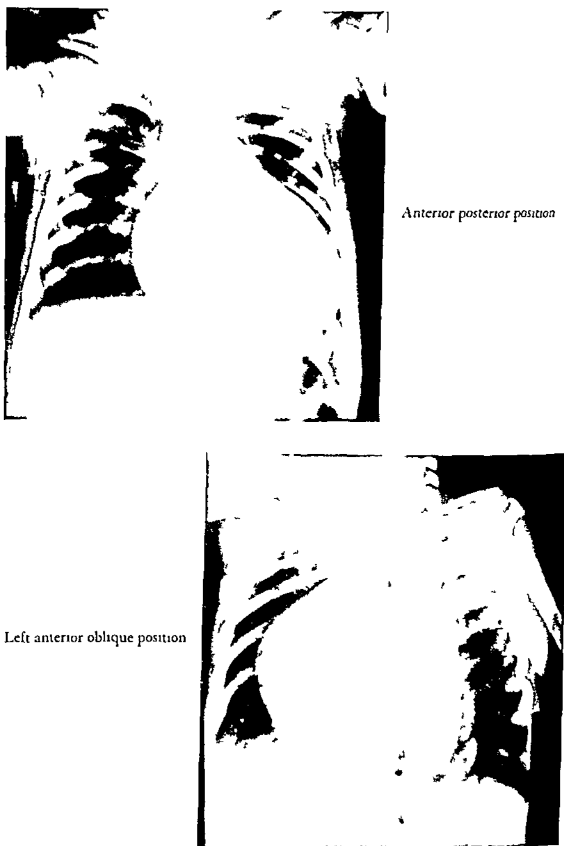


FIGURE X-11 Complete transposition of the great vessels with a high ventricular septal defect Infant

from the right ventricle and the pulmonary artery *from the* left ventricle, the aorta lies anteriorly and further to the left than *it does* in the normal heart, the pulmonary artery lies *posteriorly* and further to the right (see Figure X-1) By this *counterclockwise* rotation the pulmonary artery comes to lies behind the aorta, consequently, unless the pulmonary artery is *abnormally* large, the shadow cast by the great vessels is narrow Upon rotation of the infant into the left anterior-oblique position, the pulmonary artery, which in the anterior posterior position lies behind the aorta, comes to lie parallel to the aorta and thereby causes the shadow cast by these vessels to increase in width By placing one finger behind the other and then rotating the hand 45°, *the change in the* width of the shadow cast by the great vessels can be clearly demonstrated Owing to the patient s lack of cooperation, satisfactory roentgenograms in infancy, especially in the oblique positions, are difficult to obtain For this reason fluoroscopic examination is superior

Dilatation of the superior vena cava which is of common occurrence in cardiac failure, may cause the shadow at the base of the heart to appear abnormally wide The shadow cast by the superior vena cava always lies to the right of the sternum It can be differentiated from that of the great vessels by the *rotation* of the infant s head to the left This maneuver causes the shadow cast by the great vessels to shift to the left but never appreciably alters the shadow of the superior vena cava

Rhythmic changes in the size of the right auricle should be sought for, because when present they are of diagnostic significance This is one of the few malformations which *permit such* variations, owing to the piling up of blood on one side of the heart As the blood piles up *in the right* auricle, that chamber will gradually distend until the pressure in the right auricle becomes *sufficiently* greater than that *in the* left auricle to force open the valve of the foramen ovale Thereupon the blood will flow from the *right* auricle to the left, the right auricle will collapse, and the high pressure in the left auricle will *close the* valve covering the foramen *ovale* Thereafter the pressure in the right auricle will steadily rise until its pressure again exceeds that *in the* left auricle Then the valve will open again and the process will be repeated This causes *a* rhythmic change in the size of the *right* auricle which is independent of the heart rate This type of change is indicative of a *malformation* which causes the blood to accumulate *in the right* auricle, combined with a foramen *ovale* which is covered by a valve that is not completely sealed and permits the flow of blood *in only* one direction, namely, from right to left

Alteration in the contour of the heart associated with the growth of the patient If the nature of the concomitant malformation is such that the condition does not cause progressive cardiac enlargement, a circulatory balance may be established, and the patient may live for a number of years As previously stated, this is more likely to occur if there is pulmonary stenosis or great dilatation of the pulmonary artery than if both great vessels are of normal size If such a balance is possible, it usually occurs before the heart is enormously enlarged As the child grows and the diaphragm descends, the heart occupies a more vertical position In the anterior posterior position the contour of the heart has a concave curve at the base, owing to the posterior location of the pulmonary artery (see Figure x-12) Furthermore, in the left anterior-oblique position the shadow cast by the great vessels no longer increases in width As the heart drops down, the pulmonary artery, which lies posterior to the aorta, extends to the lungs more horizontally than it does in the normal heart The consequence is that the pulmonary artery lies at an abnormally low level and the pulmonary window becomes abnormally clear For this reason the contour of the heart in the anterior posterior position and in the oblique positions may closely resemble that of a tetralogy of Fallot



FIGURE X-12 Complete transposition of the great vessels with pulmonary stenosis Child

The vascular markings are of great diagnostic importance Most malformations which cause persistent cyanosis in early infancy are associated with a great reduction in the pulmonary blood flow and excessively clear lung fields In a complete transposition of the great vessels, the volume of the pulmonary blood flow is normal, the vascular markings in infancy are normal or slightly increased and extend from the hilar region nearly to the periphery of the lungs

In childhood the vascular markings are distinctive in the x ray film Even when there is pulmonary stenosis, the pulmonary artery is a fair sized vessel There are many small, discrete, circular shadows due to blood vessels which are viewed on end as they course from the posteriorly placed pulmonary artery to the anterior portion of the lungs Moreover, the vascular markings extend far out into the lung fields, indeed nearly to the periphery of the lungs Although the pulmonary arteries are seen to pulsate vigorously at operation, the pulsations are rarely visible upon fluoroscopy This is due in part to the fact that the pulmonary arteries lie abnormally deep within the chest, so that there is a considerable amount of lung tissue superimposed upon them, and in part because, even though the pressure within the pulmonary arteries is high, the pulse pressure in these arteries is not great Usually fluoroscopy does not reveal the details of these shadows and only the dense and numerous shadows in the hilar regions are visible Consequently the shadows cast by the small branches of pulmonary arteries viewed on end may be mistaken for those due to *extensive collateral* circulation by way of the posterior mediastinal vessels Not infrequently it is the discrepancy between the vascular shadows seen on the x ray film and those discerned upon fluoroscopy which offers the clue to correct diagnosis

In an older patient, upon slow rotation during fluoroscopic examination, it is usually possible to visualize the main branches of the pulmonary artery as they course to the lungs This may be possible in either oblique position but the *left* pulmonary artery is *generally* more readily seen in the left anterior-oblique position than in the right Indeed during fluoroscopy the lower branches of the left pulmonary artery are often visible as the patient is slowly rotated toward the left anterior-oblique position

Pulmonary stenosis renders the vascular markings even more closely similar to those of a tetralogy of Fallot Nevertheless, since the pulmonary artery arises from the left ventricle, the pulmonary stenosis is almost invariably valvular and the pulmonary artery is a fair sized vessel Consequently the vascular markings are heavier and extend further to the periphery of the lungs than in a tetralogy of Fallot So few patients who have a complete transposition of the great vessels

as a large ventricular septal defect because of failure to appreciate the significance of a femoral arterial saturation of 94 per cent Although in an Eisenmenger complex the saturation in the aorta may be the same as it is in the pulmonary artery, when the oxygen saturation is lower in the aorta than in the pulmonary artery, the possibility of a complete transposition of the great vessels should always be considered

Angiocardiography may be of aid in that it will demonstrate early opacification of the aorta and may or may not demonstrate late but extensive opacification of the pulmonary vascular bed (see Figure x-14) Unless the aorta is grossly misplaced (see Section c), angiocardiography is not of great help in the differentiation of dextroposition of the aorta from transposition of the aorta, and therefore the test is seldom of aid in the differentiation of transposition of the great vessels from a tetralogy of Fallot Occasionally in the lateral view, when both the aorta and the pulmonary artery are visualized simultaneously, the pulmonary artery can be seen to arise posterior to the aorta, thus proving that the great vessels are transposed (see Figure x-22)

DIAGNOSIS

The diagnosis is based upon the finding of a small, scrawny infant with cyanosis, rapid respirations, and a barrel shaped chest, who suffers from increasing cardiac difficulty associated with progressive cardiac enlargement, a gallop rhythm, engorgement of the liver, and congestion in the lungs When, in addition, the hands are more cyanotic than the feet, the diagnosis can be made with assurance

The x ray and fluoroscopic findings of an egg shaped heart with a narrow shadow at its base, which increases in width in the oblique view, is strong confirmatory evidence of a complete transposition of the great vessels

Children and young adults show deep cyanosis, marked clubbing of the fingers and the toes, and extreme stunting of growth The contour of the heart is similar to that of a tetralogy of Fallot The second sound over the pulmonary area is accentuated X ray findings of cardiac enlargement and a concave curve at the base of the heart to the left of the sternum, combined with dense vascular shadows which extend to the periphery of the lungs, are diagnostic of this condition

DIFFERENTIAL DIAGNOSIS

This malformation must be differentiated from a pure pulmonary stenosis, from a tetralogy of Fallot, from a single ventricle with complete transposition of

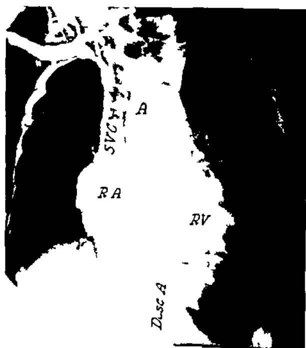




FIGURE X-14 Complete transposition of the great vessels Infant

the great vessels, and also from a truncus arteriosus with reduced pulmonary blood flow and a defective development of the right ventricle with or without tricuspid atresia

In the neonatal period the malformation may be confused with a *"pure" pulmonary stenosis* Confusion arises because in a patient with a "pure" pulmonary stenosis the foramen ovale is often functionally patent during the first few weeks of life Therefore the infant may appear cyanotic Poststenotic dilatation has not yet developed Consequently there is no fullness of the pulmonary conus In addition, in both malformations the vascular markings are more conspicuous than in other types of pulmonary stenosis Furthermore, occasionally the infant may suffer from right sided cardiac failure in the neonatal period The second sound at the base of the heart to the left of the sternum is of aid in the differentiation of the two malformations It is weak or absent in a baby with "pure" pulmonary stenosis and accentuated when there is a complete transposition of the great vessels Time is also a great aid in the differentiation of these two malformations An infant with a "pure" pulmonary stenosis usually responds to digitalis, his cyanosis disappears, and he does better than might be expected, whereas in the malformation under discussion, if the infant is in severe difficulty during the first days of life, he seldom, if ever, survives more than a few weeks

In early infancy *a tetralogy of Fallot with anatomical or functional pulmonary atresia* may be confused with a complete transposition of the great vessels combined with an intact ventricular septum and minimal communication between the two sides At birth, in both instances, cyanosis may be intense and the heart is normal in size The outstanding difference between the two malformations is that in a complete transposition of the great vessels the lung fields show normal or increased vascularity, whereas in a tetralogy of Fallot with pulmonary atresia the lung fields are phenomenally clear Further, in a complete transposition of the great vessels the heart usually enlarges rapidly Indeed, when the condition is such as to cause difficulty in the neonatal period, the enlargement can usually be detected within four to six days after birth, whereas in a tetralogy of Fallot, the heart remains small

In older children *a tetralogy of Fallot* with a severe degree of pulmonary stenosis may be confused with a complete transposition of the great vessels in which a balance is established and the heart is not greatly enlarged A tetralogy of Fallot causes less stunting of growth, there is usually left sided chest deformity and the heart is often abnormally small, the hilar shadows may appear dense but the shadows do not extend far out into the lungs In contrast to this, a complete

transposition of the great vessels causes extreme stunting of growth, the chest is usually barrel shaped, the heart is slightly enlarged, and the vascular markings extend far out to the periphery of the lungs (see Section c)

A single ventricle with transposition of the great vessels may be confused with a complete transposition of the great vessels and a normal ventricular septum, especially when the pulmonary artery is abnormally small Clinically a patient with a single *ventricle does* better than one with a complete transposition of the great vessels and two ventricles, as the single ventricle permits a good opportunity for some crossing between the venous and the arterial circulation Nevertheless, it may require cardiac catheterization or angiocardiography to determine whether or not there are two ventricles

A truncus arteriosus causes great enlargement of both ventricles Although the enlargement occurs early, it is not progressive The aortic knob is usually conspicuous If the infant lives to childhood, a continuous murmur is usually audible over some portion of the lung

Defective development of the right ventricle with tricuspid atresia is characterized by clear lung fields and electrocardiographic evidence of a left axis deviation and left ventricular hypertrophy

Defective development of the right ventricle with pulmonary stenosis and an intact ventricular septum causes cardiac enlargement and great reduction in the pulmonary blood flow The second sound at the base of the heart to the left of the sternum is diminished and upon x ray the vascular markings are decreased Cardiac catheterization will show a high pressure in the right ventricle but the aorta cannot be entered Angiocardiography shows that the pulmonary artery, not the aorta arises from the right ventricle

TREATMENT

Digitalis may prolong the life of the infant

The inhalation of oxygen may be of temporary benefit

Surgical treatment has not yet been perfected The creation of an auricular septal defect, as developed by Blalock [12] has been of benefit to some patients The best results have been seen in children over two years of age These patients have shown an increase in their exercise tolerance and over a period of years some reduction has occurred in the red blood cell count, the amount of available hemoglobin and the hematocrit level Thus although the immediate results are seldom dramatic the long time results have been better than anticipated Nevertheless, this operation does not relieve the pulmonary hypertension

Baffes[13] has developed an operation for the partial transposition of the pulmonary veins to compensate for the transposition of the great vessels In this operation the right pulmonary veins are transplanted into the right auricle and a graft is inserted between the inferior vena cava and the left auricle The improvement after this operation is dramatic Initially the best results were obtained in children over two years of age, recently excellent results have also been obtained in young infants

Dr Henry Bahnson has performed one operation in which he altered the position of the auricular septum, so that the pulmonary veins drained into the right auricle and the inferior vena cava into the left auricle This operation may prove to be of great benefit

PROGNOSIS

The prognosis is poor Most infants, when the auricular and ventricular septa are closed, die of anoxemia during the first weeks of life, some infants live for a few months but the condition usually leads to progressive cardiac enlargement These infants usually die of anoxemia or cardiac failure between six and fifteen months of age Only occasionally is a balance established which permits the patient to live until childhood or early adult life

Operation definitely improves the prognosis As of 1960 the greatest improvement has resulted from the Baffes operation

SUMMARY

A complete transposition of the great vessels means that the aorta arises from the right ventricle and the pulmonary artery from the left ventricle It is a relatively common malformation, as it places no strain on the fetal circulation

After birth the blood is pumped around and around the systemic circulation and around and around the pulmonary circulation Great difficulty is encountered in the crossing of the two circulations The larger the shunt, the less intense is the cyanosis Nevertheless, the blood which is shunted from one side of the heart to the other tends to pile up on the side to which it is shunted In order to reverse the shunt, the relative pressure in the two circulations must be reversed This means that the pulmonary pressure must be approximately the same as the systemic pressure Pulmonary hypertension is the rule

Cyanosis may not be apparent during the first weeks of life, sooner or later it always develops and eventually becomes intense The distribution of the cyanosis may give a clue to the diagnosis

Clubbing develops early

Polycythemia steadily increases and may become extreme

An infant with this malformation usually suffers from severe anoxemia and may suffer from episodes of loss of consciousness but attacks of paroxysmal dyspnea are rare Respirations are rapid and shallow The chest is generally barrel shaped Difficulty in feeding and failure to gain are common complaints Stunting of growth may be extreme The exercise tolerance is always limited The child may squat when tired

The heart usually undergoes progressive enlargement As it enlarges, it develops a characteristic contour Both ventricles are always markedly enlarged There is absence of fullness of the pulmonary conus In infancy the shadow cast by the great vessels is narrow in the anterior posterior position and upon rotation of the patient into the left anterior-oblique position the shadow cast by the great vessels increases in width Inasmuch as there is adequate circulation to the lungs, the infant is likely to suffer from pulmonary congestion Death results from anoxemia or from cardiac failure with congestion in the lungs and edema of the extremities

Occasionally the malformation is compatible with life for a number of years Under such circumstances the contour of the heart and both the physical and the physiological findings closely resemble a tetralogy of Fallot The two features which differentiate this malformation from that of a tetralogy of Fallot are the accentuation of the second sound at the base of the heart to the left of the sternum and the density of the vascular markings which extend from the hilar region to the periphery of the lungs

The electrocardiogram ordinarily shows a right axis deviation and evidence of hypertrophy of both ventricles

The circulation time (arm to tongue) is abnormally short

The oxygen saturation of the arterial blood is abnormally low

Cardiac catheterization is of no great aid in the diagnosis of a complete transposition of the great vessels If the pulmonary artery is entered, the oxygen saturation of the blood in the pulmonary artery is always found to be higher than that in the femoral artery thus catheterization may occasionally give a clue to the existence of a transposition of the great vessels Cardiac catheterization may also aid in the diagnosis of the nature of the concomitant malformation

Angiocardiography shows early dense opacification of the aorta In the lateral series of films it may be possible to see that the aorta lies anterior to the pulmonary artery

The diagnosis of a complete transposition of the great vessels is based upon the finding of persistent cyanosis, increasing cardiac enlargement, and cardiac failure with engorgement of the liver and congestion of the lungs When, in addition, the hands are more cyanotic than the feet, the diagnosis can be made with assurance The diagnosis is further confirmed by the x ray evidence of a concavity at the base of the heart, absence of the shadow cast by the pulmonary conus, a narrow pedicle in the anterior-posterior position, which increases in width in the left anterior-oblique position, and dense hilar shadows with extensive vascular markings

The malformation requires differentiation from a 'pure" pulmonary stenosis, from a tetralogy of Fallot, from a single ventricle, and occasionally from a truncus arteriosus with reduced pulmonary blood flow

The creation of an auricular defect may aid in the crossing of the two circulations but does not lessen the pulmonary hypertension The Baffes operation offers greater hope for improvement of the circulation

The prognosis is poor Most infants with this malformation die between six and fifteen months of age Without operation only rarely does a patient live to adolescence or early adult life

B *Complete Transposition of the Great Vessels Combined with Enormous Dilatation of the Pulmonary Artery*

NATURE OF THE MALFORMATION

In this malformation the aorta, which arises from the right ventricle, is usually of normal size and the pulmonary artery, which arises from the left ventricle, is huge When the pulmonary artery is tremendously enlarged, it frequently over rides the ventricular septum, under such circumstances a high ventricular septal defect is an integral part of the malformation (see Figures X-15 and 16) It is, however, important to appreciate that a complete transposition of the great vessels combined with enormous dilatation of the pulmonary artery may occur in combination with only a small ventricular septal defect or even an intact ventricular septum

COURSE OF THE CIRCULATION

The course of the circulation differs from that in other cases of transposition of the great vessels only if the pulmonary artery arises in part from the right

RA
Pulmonary a.
RA
L auricle
Ventricular septal defect
Mitral v
Tricuspid v
R ventricle
L ventricle

FIGURE X-15 Complete transposition of the great vessels with an enormously dilated pulmonary artery

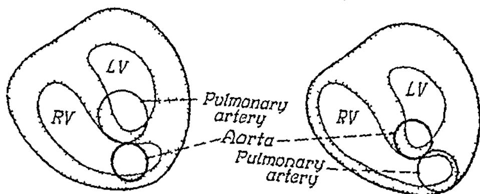


FIGURE X-16 Complete transposition of the great vessels with a dilated pulmonary artery and normal heart

ventricle. Under such circumstances the blood in the right ventricle is pumped out through the aorta to the systemic circulation and through the pulmonary artery to the lungs. The blood which is directed to the systemic circulation is returned by the superior and inferior venae cavae to the right auricle and that which goes to the lungs is returned by the pulmonary veins to the left auricle and thence to the left ventricle. Most of the blood from the left ventricle is pumped out into the pulmonary artery. When the pulmonary artery over-rides the ventricular septum, some venous blood from the right ventricle is readily shunted to the lungs, and consequently an increased volume of blood is returned to the left side of the heart, which aids in the shunting of oxygenated blood across to the right side of the heart. Thus some crossing of the circulation can occur, as shown in Diagram x–5. For this reason, it is not surprising to find that this combination of anomalies is frequently more compatible with life than when both great vessels are of normal size and there is no over-riding of the pulmonary artery.

PHYSIOLOGY OF THE MALFORMATION

The physiology of this malformation is essentially the same as that when both great vessels are of normal size, except that the volume of the pulmonary blood flow is greater. When the pulmonary artery over-rides the ventricular septum, the condition is physiologically and functionally similar to that of a Taussig-Bing malformation, as the pulmonary artery is biventricular in origin (see Chapter xi). Nevertheless, when the pulmonary artery lies posterior to the aorta, it receives mainly oxygenated blood from the left ventricle, whereas when it lies anterior and to the left of the aorta it receives more venous blood. As in all cases of complete transposition of the great vessels, it is the relative pressure in the two circulations which regulates the volume of the shunt. Only when pulmonary pressure is greater than systemic pressure is oxygenated blood directed to the aorta. It follows that, regardless of whether or not the pulmonary artery over-rides the ventricular septum, there is always pulmonary hypertension.

CLINICAL FINDINGS

Cyanosis is usually present at birth. Inasmuch as the aorta arises from the right ventricle, cyanosis almost invariably appears early and eventually becomes intense. Owing to the fact that the shunt is within the heart, the cyanosis is of uniform distribution.

Clubbing of the extremities appears at an early age and may become extreme

Polycythemia increases with age and eventually becomes extreme As the patient grows older the usual changes in the clotting mechanism occur In late childhood or early adolescence, petechiae and blotchy purpuric eruption frequently occur

Respirations are rapid and shallow, and the chest becomes barrel shaped These infants may suffer from prolonged periods of unconsciousness due to extreme anoxemia

Difficulty in feeding, failure to gain and stunting of growth occur, as in all cases of complete transposition of the great vessels

Exercise tolerance is limited Children may squat when tired

Pulmonary congestion is common, owing to the excessive pulmonary blood flow Furthermore, the excessive pulmonary circulation renders the individual very susceptible to bronchitis and pneumonia Indeed, it is frequently difficult to determine whether the pulmonary congestion is due to cardiac failure or to infection

Hepatomegaly is the rule Even though a balance is established, there is usually slight to moderate engorgement of the liver

CARDIAC FINDINGS

The heart is always slightly enlarged A balance is established early in life and therefore the heart is not enormously enlarged

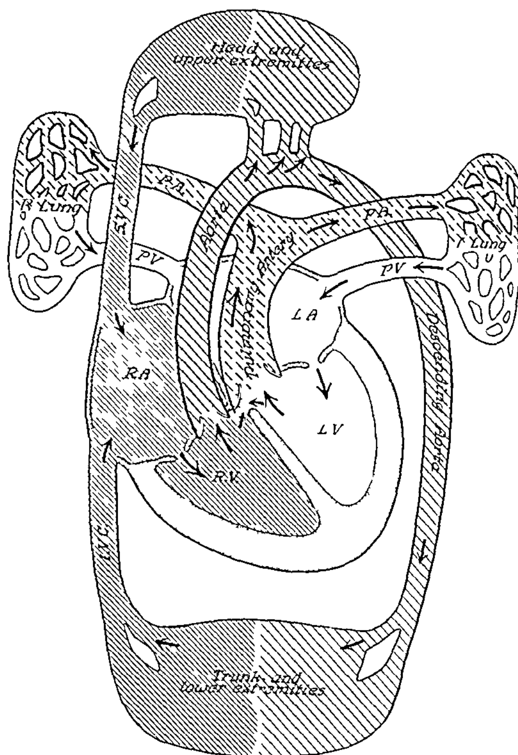
The pulmonic second sound is usually accentuated and may be *reduplicated*

A systolic murmur is the rule Ordinarily the *murmur* is not very harsh Frequently a *blurred third heart sound* or a *mid diastolic murmur* is audible just inside the apex

X RAY AND FLUOROSCOPIC FINDINGS

The heart is usually slightly enlarged and there is a concave *curve* at its base to the left of the sternum In early infancy the vascular markings are increased but may not be conspicuous (see Figure x–17) Nevertheless, owing to the great dilatation of the pulmonary artery, as the patient grows, the hilar markings become exaggerated (see Figure x–18) Usually there are large, blotchy hilar shadows which may or may not pulsate The pulsations in these vessels are often less conspicuous than would be expected from their size, because, as previously mentioned, although the pulmonary blood flow is excessive, both the systolic and the diastolic pressure in the pulmonary artery are high, and consequently the pulse pressure in the pulmonary artery is not wide In older children, however, a hilar

DIAGRAM X-5



Arterial blood (fully saturated)

Venous and arterial blood
Cyanosis visible

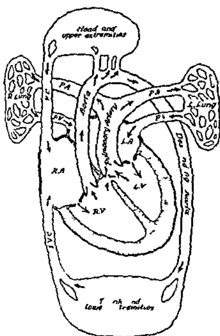
Small admixture of venous blood
No visible cyanosis

Venous blood

Diagram X-5

Complete transposition of the great vessels with a greatly dilated pulmonary artery

In this malformation a complete transposition of the great vessels is combined with a greatly dilated pulmonary artery, which frequently over rides the ventricular septum and thus receives blood from both ventricles In some instances, however, the pulmonary artery does not over ride the ventricular septum (see insert)



The blood from the right auricle flows into the right ventricle. Inasmuch as the aorta arises from the right ventricle, most of the blood in the right ventricle is pumped directly out through the aorta to the body and returned by the superior and inferior venae cavae to the right auricle, thence it again flows into the right ventricle. If the large pulmonary artery arises in part from the right ventricle some blood is pumped into the pulmonary artery, or if the pressure is lower in the lungs, blood is shunted from the right side to the left All of the blood so shunted and most of the blood from the left ventricle is pumped out through the pulmonary artery to the lungs, where it is fully oxygenated and returned by the pulmonary veins to the left auricle, thence it flows to the left ventricle. Again most of the blood from the left ventricle re-circulates through the lungs When however, the pressure on the left side exceeds that on the right side, some oxygenated blood from the left side will be shunted to the right side There it will be mixed with the venous blood in the right ventricle and will be pumped out through the aorta to the body and returned by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

In this malformation as in all cases of complete transposition of the great vessels except for those with pulmonary stenosis it is the relative pressure in the two circulations which regulates the volume of the shunt Hence there is always severe pulmonary hypertension

Clinical diagnosis The patient shows deep persistent cyanosis Murmurs are not significant The great size of the pulmonary artery and the relatively easy shunting of blood frequently enable a balance to be established and the heart ceases to enlarge The condition may be compatible with life for a number of years The x ray shows slight cardiac enlargement a concave curve at the base of the heart to the left of the sternum, and extremely vascular lung fields The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy



At one week

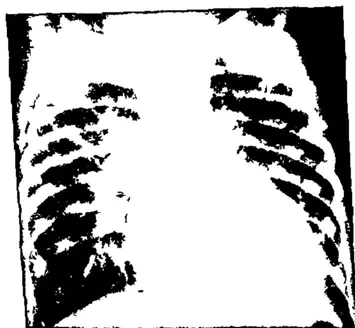


At two weeks

FIGURE X–17 Complete transposition of the great vessels with a dilated pulmonary artery Infant



At one and one quarter years



At two and one-quarter years

Figure x-18 Complete transposition of the great vessels with a dilated pulmonary artery Child

dance is the rule Regardless of the presence or absence of a hilar dance, conspicuous vascular shadows indicate a huge pulmonary artery, when this is combined with a concave curve at the base of the heart, as shown in Figure X–19, it is diagnostic of a large posteriorly placed pulmonary artery Hence these findings are almost pathognomonic of a complete transposition of the great vessels combined with dilatation of the pulmonary artery In rare instances the main pulmonary artery is so greatly dilated that it gives the appearance of fullness of the pulmonary conus (see Figure X–20)

In the left anterior oblique position both ventricles are seen to be enlarged and the huge pulmonary artery fills the pulmonary window

In the right anterior-oblique position the left auricle is seen to be of normal size

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is similar to that in other cases of transposition of the great vessels There is a right axis deviation in the standard leads and evidence of right ventricular hypertrophy in the unipolar precordial leads

SPECIAL TESTS

The circulation time (arm to tongue) is abnormally short

The oxygen saturation of the arterial blood is abnormally low and falls still further with exercise

Cardiac catheterization is usually not necessary for diagnosis but is of aid in the determination of the structure of the auricular septum and whether or not there is a single ventricle When the pulmonary artery over rides the ventricular septum it may be possible to catheterize the pulmonary artery If the pulmonary artery is entered, the oxygen content of the sample of blood in the pulmonary artery will be found to be higher than that in the aorta, indeed, the blood in the pulmonary artery is usually almost fully saturated The pressure in the pulmonary artery is always abnormally high Thus the catheterization findings are closely similar to those of a Taussig Bing malformation Indeed, it is the course which the catheter takes as it enters the pulmonary artery which indicates whether the pulmonary artery occupies its normal position or lies posterior to the aorta

Angiocardiography is almost always disappointing because the dye is dissipated from the right ventricle to both circulations and also because the large size of the pulmonary vessels, combined with the large volume of blood directed to

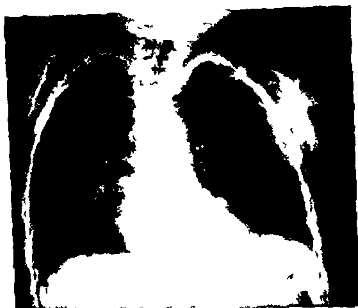


FIGURE X-19 Complete transposition of the great vessels with a greatly dilated pulmonary artery Child

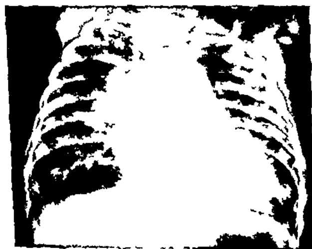


FIGURE X-20 Complete transposition of the great vessels with a greatly dilated pulmonary artery Infant

dance is the rule Regardless of the presence or absence of a hilar dance, conspicuous vascular shadows indicate a huge pulmonary artery, when this is combined with a concave curve at the base of the heart, as shown in Figure X-19, it is diagnostic of a large posteriorly placed pulmonary artery Hence these findings are almost pathognomonic of a complete transposition of the great vessels combined with dilatation of the pulmonary artery In rare instances the main pulmonary artery is so greatly dilated that it gives the appearance of fullness of the pulmonary conus (see Figure X-20)

In the left anterior oblique position both ventricles are seen to be enlarged and the huge pulmonary artery fills the pulmonary window

In the right anterior-oblique position the left auricle is seen to be of normal size

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is similar to that in other cases of transposition of the great vessels There is a right axis deviation in the standard leads and evidence of right ventricular hypertrophy in the unipolar precordial leads

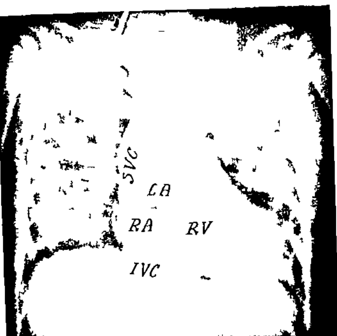
SPECIAL TESTS

The circulation time (arm to tongue) is abnormally short

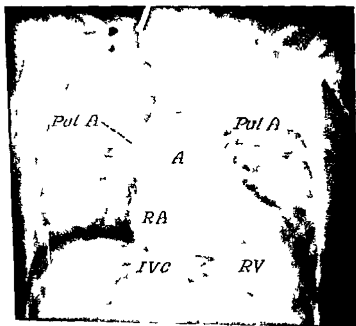
The oxygen saturation of the arterial blood is abnormally low and falls still further with exercise

Cardiac catheterization is usually not necessary for diagnosis but is of aid in the determination of the structure of the auricular septum and whether or not there is a single ventricle When the pulmonary artery over rides the ventricular septum it may be possible to catheterize the pulmonary artery If the pulmonary artery is entered, the oxygen content of the sample of blood in the pulmonary artery will be found to be higher than that in the aorta, indeed, the blood in the pulmonary artery is usually almost fully saturated The pressure in the pulmonary artery is always abnormally high Thus the catheterization findings are closely similar to those of a Taussig Bing malformation Indeed, it is the course which the catheter takes as it enters the pulmonary artery which indicates whether the pulmonary artery occupies its normal position or lies posterior to the aorta

Angiocardiography is almost always disappointing because the dye is dissipated from the right ventricle to both circulations and also because the large size of the pulmonary vessels, combined with the large volume of blood directed to



Dye in the superior vena cava, the right auricle and the right ventricle



Dye immediately passes to the aorta and the pulmonary artery

FIGURE X-21 Complete transposition of the great vessels with a posteriorly placed dilated pulmonary artery Child

the pulmonary artery from the left ventricle, causes excessive dilution of the dye in the lungs (see Figure x–21) Films taken with the patient in the lateral position, however, confirm the fact that the pulmonary artery arises far posteriorly (see Figure x–22)

DIAGNOSIS

The diagnosis is suggested by the deep, persistent cyanosis and clubbing in a patient with evidence of a poorly functioning heart and slight enlargement of the liver and congestion in the lungs It is confirmed by the x ray and by fluoroscopic findings of large, blotchy hilar shadows which may or may not show a conspicuous hilar dance, combined with a concave curve at the base of the heart to the left of the sternum which shows that the pulmonary artery is posteriorly placed

DIFFERENTIAL DIAGNOSIS

The two conditions with which this malformation are most frequently confused are (1) a single ventricle combined with a complete transposition of the great vessels and enormous dilatation of the pulmonary artery and (2) a Taussig-Bing malformation

A single ventricle combined with complete transposition of the great vessels and enormous dilatation of the pulmonary artery may closely resemble the malformation under discussion When the great vessels are completely transposed, a patient with a single ventricle usually does better than a patient with two ventricles, as there is a better admixture of venous and arterial blood Cardiac catheterization is usually necessary to differentiate the two conditions It is the difference in the oxygen content of the blood in the common ventricle in comparison with that in the right auricle which gives the clue to the correct diagnosis When there are two ventricles, the increase in the oxygen content of the blood in the right ventricle is comparatively slight, whereas, if there is but a single ventricle, there is usually an increase of 5 volumes per cent (see Chapter xv)

If there is a defect in the auricular septum, it will cause an increase in the oxygen content of the blood in the right auricle in comparison with that in the superior vena cava Needless to say, this will alter the percentage of increase in the oxygen content of the blood in the ventricle in comparison with that in the right auricle

A Taussig Bing malformation may occasionally be confused with the malformation under discussion, because in certain instances the contours of the heart are similar In a complete transposition of the great vessels the greatly dilated

(compare Figures X–22 and XI–6) Functionally the two malformations are closely similar but accurate differentiation is important if surgery is contemplated

TREATMENT

Patients with an intact auricular septum can be helped either by a Baffes operation or by the creation of an auricular septal defect, provided that the oxygen content of the blood in the pulmonary artery is significantly greater than that in the aorta If, however, the oxygen content of the blood in the aorta and in the pulmonary artery are approximately the same, as in the case of a single ventricle, there is little to gain from operation

PROGNOSIS

The prognosis is usually better when the pulmonary artery is abnormally large than when both great vessels are of normal size A number of patients with the former condition live until late childhood and some live to adult life

During infancy and childhood the increased pressure in the lesser circulation increases the volume of oxygenated blood shunted into the aorta Nevertheless, the condition leads to progressive injury to the pulmonary vascular bed and eventually it leads to a reduction in the circulation to the lungs and hence reduces the volume of the effective flow These changes cause the patient to become progressively more incapacitated as he reaches adult life

SUMMARY

Complete transposition of the great vessels combined with enormous dilatation of the pulmonary artery is usually associated with a ventricular septal defect and slight over riding of the pulmonary artery Under these circumstances there is better opportunity for the crossing of the two circulations than in other types of complete transposition of the great vessels Cyanosis becomes apparent at an early age and is of uniform distribution Polycythemia and clubbing usually develop during the first two years of life Respirations are rapid and shallow and the infant s chest becomes barrel shaped The liver becomes enlarged The heart is slightly enlarged A gallop rhythm or a pathological third heart sound is audible at the apex

The x ray and fluoroscopic findings are distinctive in that there is evidence of a huge pulmonary artery combined with a concave curve at the base of the heart to the left of the sternum

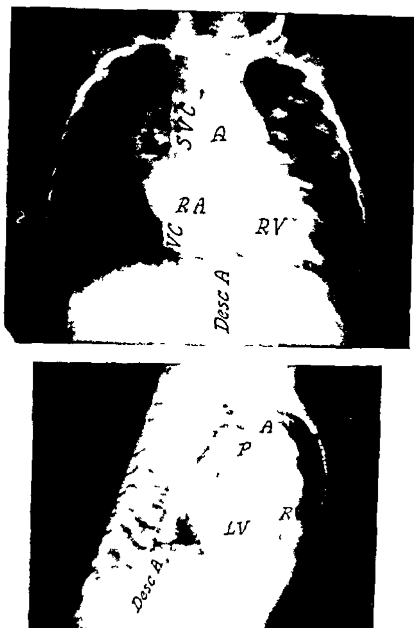


Figure x-22 Complete transposition of the great vessels with an abnormally large pulmonary artery Child

pulmonary artery may cast a shadow to the left of the aorta and thus cause fullness of the pulmonary conus instead of the usual concavity (compare Figures x-20 and xi-4) In the author's experience the vascularity of the lungs is usually more exaggerated when the pulmonary artery arises from the left ventricle Angiocardiography will help to differentiate the two conditions In a Taussig Bing malformation the anterior posterior series of films shows simultaneous opacification of both great vessels as they lie side by side When the great vessels are transposed the pulmonary artery will be seen to lie posterior to the aorta

(compare Figures X-22 and XI-6) Functionally the two malformations are closely similar but accurate differentiation is important if surgery is contemplated

TREATMENT

Patients with an intact auricular septum can be helped either by a Baffes operation or by the creation of an auricular septal defect, provided that the oxygen content of the blood in the pulmonary artery is significantly greater than that in the aorta If, however, the oxygen content of the blood in the aorta and in the pulmonary artery are approximately the same, as in the case of a single ventricle, there is little to gain from operation

PROGNOSIS

The prognosis is usually better when the pulmonary artery is abnormally large than when both great vessels are of normal size A number of patients with the former condition live until late childhood and some live to adult life

During infancy and childhood the increased pressure in the lesser circulation increases the volume of oxygenated blood shunted into the aorta Nevertheless, the condition leads to progressive injury to the pulmonary vascular bed and eventually it leads to a reduction in the circulation to the lungs and hence reduces the volume of the effective flow These changes cause the patient to become progressively more incapacitated as he reaches adult life

SUMMARY

Complete transposition of the great vessels combined with enormous dilatation of the pulmonary artery is usually associated with a ventricular septal defect and slight over riding of the pulmonary artery Under these circumstances there is better opportunity for the crossing of the two circulations than in other types of complete transposition of the great vessels Cyanosis becomes apparent at an early age and is of uniform distribution Polycythemia and clubbing usually develop during the first two years of life Respirations are rapid and shallow and the infant s chest becomes barrel shaped The liver becomes enlarged The heart is slightly enlarged A gallop rhythm or a pathological third heart sound is audible at the apex

The x ray and fluoroscopic findings are distinctive in that there is evidence of a huge pulmonary artery combined with a concave curve at the base of the heart to the left of the sternum

The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy Cardiac catheterization is often necessary to differentiate this malformation from a single ventricle with a complete transposition of the great vessels Angiocardiography is occasionally necessary to differentiate the condition from a Taussig Bing malformation Surgical correction is both difficult and unsatisfactory Nevertheless, a Baffes procedure or the creation of an auricular septal defect is of value if the oxygen content of the blood in the aorta is appreciably lower than that in the pulmonary artery

The prognosis is guarded Although the condition may be compatible with life for ten to twenty years, most persons with this malformation are severely handicapped and die at an early age

C *Complete Transposition of the Great Vessels Combined with Pulmonary Stenosis or Atresia*

Complete transposition of the great vessels may occur in combination with pulmonary stenosis or atresia In both instances the pulmonary obstruction protects the lungs but it also greatly reduces the volume of the pulmonary circulation When there is pulmonary atresia, the pulmonary circulation is so poor that the condition is not long compatible with life

When pulmonary stenosis is combined with a complete transposition of the great vessels, the pressure in the pulmonary artery is low Although the circulation is far more nearly adequate than when there is pulmonary atresia, the reduction in the volume of the pulmonary blood flow is far greater than in other types of complete transposition of the great vessels Indeed, the clinical picture more closely resembles that of a tetralogy of Fallot with a severe pulmonary stenosis than it does other types of complete transposition of the great vessels

NATURE OF THE MALFORMATION

As in most cases of complete transposition of the great vessels, the aorta usually arises from the mid portion of the right ventricle and the pulmonary artery lies directly posterior to it

When there is pulmonary atresia, the only way for the blood to reach the lungs is through the ductus arteriosus or by the collateral circulation The pulmonary blood flow is very meager Little blood is returned to the left auricle, hence only a small volume of blood is pumped out from the left ventricle, the left ventricle remains small, the right ventricle, which pumps the blood to the systemic circulation, becomes hypertrophied (see Figure X-23)

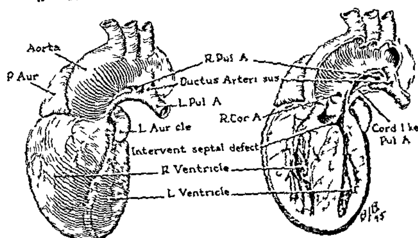


FIGURE X-23 Complete transposition of the great vessels combined with pulmonary atresia and a high ventricular septal defect

When there is pulmonary stenosis, more blood reaches the lungs and more blood is returned to the left auricle and the left ventricle The pulmonary stenosis, however, renders it difficult for the blood to be expelled from the left ventricle, therefore the left ventricle becomes hypertrophied When the pulmonary artery arises from the left ventricle, the pulmonary stenosis is almost always valvular in type, as there is no infundibular chamber from which it can arise The right ventricle pumps the blood to the systemic circulation, hence it, too, is hypertrophied Figure x-24 shows a complete transposition of the great vessels combined with a valvular pulmonary stenosis in which there is a moderately large ventricular septal defect Such is the usual finding in this malformation

In some instances the aorta is further transposed and occupies the position of the normal pulmonary artery Under such circumstances, the ascending aorta arches boldly to the left and turns to the right and may descend on the left or the right of the vertebral column

As in all cases of complete transposition of the great vessels, there is always some additional malformation A ventricular septal defect is common, occasionally there is only an auricular septal defect

COURSE OF THE CIRCULATION

During fetal life the course of the circulation is altered both by the abnormal position of the aorta and by the pulmonary stenosis or atresia Most of the blood in the right auricle flows into the right ventricle and is pumped out through the aorta to the body of the fetus and is returned by the superior vena cava and the

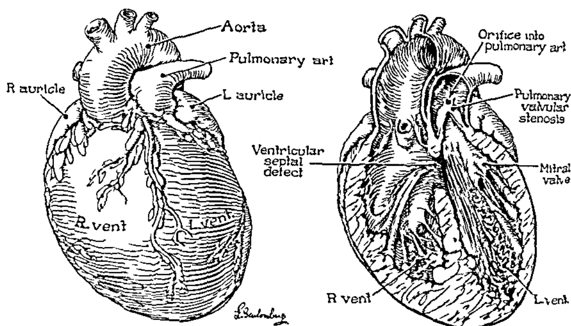


FIGURE X-24 Complete transposition of the great vessels combined with valvular pulmonary stenosis and a ventricular septal defect

inferior vena cava to the right auricle Some of the blood from the right auricle flows into the left auricle and thence to the left ventricle Here the blood encounters difficulty in reaching the aorta, hence the pressure on the left side of the heart tends to rise and this, in turn, tends to lessen the volume of blood which flows from the right auricle to the left The body of the fetus receives adequate circulation from the aorta Even though the pulmonary artery is atretic at its base, the ductus arteriosus and the main branches of the pulmonary artery develop normally and the circulation to the lungs is established from the aorta through the ductus arteriosus to the pulmonary artery (see Figure X-25) The fetal circulation is more easily established when there is pulmonary stenosis (see Figure X-26) than when there is pulmonary atresia

After birth the course of the circulation is essentially the same The expansion of the lungs lowers the pulmonary pressure and increases the volume of blood which flows through the ductus arteriosus to the lungs, where it is oxygenated The oxygenated blood is returned to the left auricle The valve covering the foramen ovale tends to close The blood from the left auricle is directed into the left ventricle Inasmuch as no vessel arises from the left ventricle, blood must be pumped from the left ventricle through the septal defect into the aorta The blood which is pumped into the aorta is returned in the normal manner by the superior vena cava and inferior vena cava to the right auricle and thence it

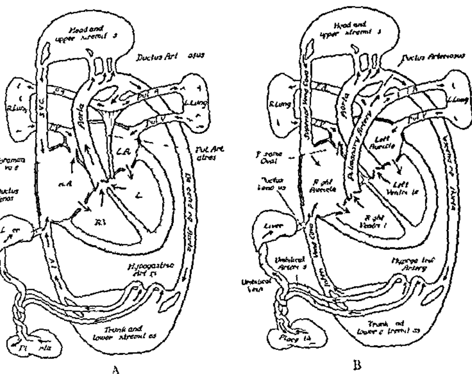


FIGURE X-25 Fetal circulation (A) Complete transposition of the great vessels combined with pulmonary atresia and (B) normal heart

is again pumped out to the systemic circulation The course of the circulation is shown in Diagram X-6

When there is pulmonary stenosis the circulation to the lungs is slightly more adequate as some oxygenated blood from the left ventricle is pumped through the stenosed artery to the lungs through which it circulates and is returned to the left auricle All the blood which is pumped out from the right side of the heart into the aorta is returned by the superior and inferior venae cavae to the right auricle and the right ventricle As in all cases of complete transposition of the great vessels, some crossing of the two circulations must occur There must be either an auricular or a ventricular septal defect or an anomaly of the venous return In the malformation under discussion the pulmonary stenosis renders it difficult for the left ventricle to empty itself The pressure within the left ventricle is increased When the pressure in the left ventricle exceeds that in the right ventricle oxygenated blood is directed to the systemic circulation through the ventricular septal defect When the pressure is greater on the right side of the heart, some venous blood is directed through the defect into the stenosed pulmo-

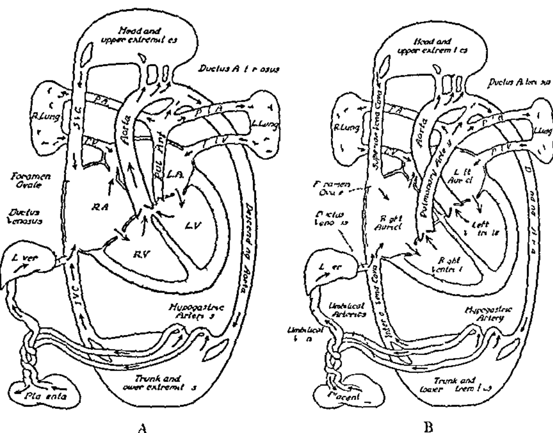


FIGURE X-26 Fetal circulation (A) Complete transposition of the great vessels combined with pulmonary stenosis and (B) normal heart

nary artery and thence to the lungs The course of the circulation is shown in Diagram X-7

When there is an auricular septal defect and no ventricular septal defect, the pressure in the left ventricle will continue to rise until the pressure in the left auricle exceeds that of the right auricle and thereupon a left to-right shunt will be established The volume of the shunt will, however, be small and the effective flow will be extremely low The course of the circulation is shown in Diagram X-8

PHYSIOLOGY OF THE MALFORMATION

Inasmuch as the aorta arises from the right ventricle, the systolic pressure in the right ventricle is the same as that in the systemic circulation Furthermore, since the only way for blood to escape from the left ventricle is through the ste nosed pulmonary artery or through the septal defect to the right ventricle, the pressure in the left ventricle is approximately the same as that in the right ven

tricle Indeed, it is the relative pressure in the two ventricles which regulates both the volume and the direction of the shunt The pulmonary stenosis or atresia, however, protects the lungs and the pressure in the pulmonary artery is low

When there is pulmonary atresia, the entire circulation to the lungs is by way of the ductus arteriosus Hence, only if the collateral circulation develops with great rapidity, is the baby able to survive the closure of the ductus arteriosus

When there is pulmonary stenosis, the pulmonary blood flow is greater than when the pulmonary artery is atretic Nevertheless, there is great difficulty in the direction of venous blood to the lungs and of arterial blood to the systemic circulation The effective flow is very low

The difficulty in the ejection of blood from the left ventricle raises the pressure in that chamber Indeed the pressure in the left ventricle must equal or exceed the pressure in the right ventricle in order to direct oxygenated blood to the systemic circulation When the pressure in the left ventricle is lower than that in the right ventricle, some blood from the right ventricle will be pumped into the left ventricle this is the main pathway by which venous blood can be directed to the lungs after the closure of the ductus arteriosus

If the ventricular septum is intact the pressure in the left ventricle will rise still further this in turn will raise the pressure in the left auricle When the pressure in the left auricle exceeds that in the right, a left to-right shunt will be established at the auricular level Over a period of years the collateral circulation develops which aids in the direction of venous blood to the lungs

CLINICAL FINDINGS

During infancy the clinical findings are closely similar to those of a tetralogy of Fallot with severe pulmonary stenosis

Cyanosis is always intense and usually dates from birth It is of uniform distribution

Attacks of paroxysmal dyspnea occur early in life as the ductus arteriosus undergoes obliteration Infants with pulmonary atresia frequently suffer from episodes of loss of consciousness and seldom survive the closure of the ductus arteriosus When, however, there is pulmonary stenosis the condition may be compatible with life for many years

Clubbing develops early and becomes pronounced

Polycythemia develops rapidly The red blood cell count may reach 11 million cells per cu mm , and the amount of available hemoglobin will be proportionately increased The hematocrit reading may be above 90 per cent Over a

Diagram x–6

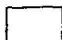
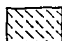
	Arterial blood (fully saturated)		Venous and arterial blood Cyanosis visible
	Small admixture of venous blood No visible cyanosis		Venous blood

Diagram X-6

Complete transposition of the great vessels pulmonary atresia a high ventricular septal defect and a patent ductus arteriosus

In this malformation the aorta arises primarily from the right ventricle and the pulmonary artery is atretic hence it makes no difference whether the pulmonary artery arises from the right or the left ventricle

The blood from the right auricle flows into the right ventricle and is pumped out through the aorta to the body The blood in the systemic circulation is returned in the normal fashion by the superior vena cava and the inferior vena cava to the *right auricle* and thence to the right ventricle Since there is pulmonary atresia, the only way for the blood to reach the lungs is through the ductus arteriosus to the pulmonary artery The blood which goes to the lungs is returned in the normal manner by the pulmonary veins to the left auricle and thence to the left ventricle. The blood from the left ventricle is pumped out into the aorta Inasmuch as the aorta arises mainly from the right ventricle there is difficulty in the expulsion of blood from the left ventricle If the duration of life is sufficiently long the left ventricle may be hypertrophied, usually the left ventricle is small The right ventricle does the main work it is hypertrophied

Clinical diagnosis The heart is enlarged especially the right ventricle In the anterior posterior position there is no fullness of the pulmonary conus and in the left anterior-oblique position the pulmonary window is wide and clear

In this malformation there is not only difficulty in the direction of the blood to the lungs for oxygenation but because the aorta arises primarily from the right ventricle, there is also difficulty in the direction of the oxygenated blood from the left ventricle into the aorta The body receives mainly venous blood from the right ventricle consequently cyanosis and dyspnea are intense The condition is not long compatible with life

Diagram x–7

Head and upper extremities

R Lung

S.V.C.

P.A.

PV

Aorta

Pulmonary Art.

Pul. A.

Pul. V.

L Lung

L.A.

R.A.

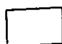
L.V.

R.V.

Descending Aorta

I.V.C.

Trunk and lower extremities



Arterial blood (fully saturated)



Small admixture of venous blood
No visible cyanosis



Venous and arterial blood
Cyanosis visible



Venous blood

Diagram X–7

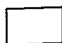
Complete transposition of the great vessels pulmonary stenosis a high ventricular septal defect and a patent foramen ovale

In this malformation there is complete transposition of the great vessels combined with pulmonary stenosis and a high ventricular septal defect The foramen ovale is frequently held open by the high pressure in the right auricle it permits the flow of blood in one direction only, namely from right to left

The blood from the right auricle flows into the right ventricle, is pumped out by way of the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle and thence to the right ventricle The blood from the left auricle flows into the left ventricle and is pumped out by way of the pulmonary artery to the lungs whence it is returned by the pulmonary veins to the left auricle The only possible way for a crossing of the two circulations is either from the right auricle through the foramen ovale to the left auricle or from either one of the ventricles through the high ventricular septal defect into the aorta or the pulmonary artery As in all cases of complete transposition of the great vessels the blood shunted from one side to the other is returned to the side to which it was shunted, it thereby raises the pressure on that side and lowers the pressure on the side from which it was shunted Thus blood shunted from the left ventricle into the aorta is returned to the right auricle and the right ventricle As the pressure in the right ventricle rises blood can be shunted through the ventricular septal defect into the stenosed pulmonary artery Nevertheless, inasmuch as there is pulmonary stenosis little blood reaches the lungs The pressure in the right ventricle rises and subsequently the pressure in the right auricle also rises The valve covering the foramen ovale is forced open and the blood is shunted from the right auricle to the left auricle thence to the left ventricle, and out by way of the pulmonary artery and the aorta Owing to the small size of the pulmonary artery, there is a greater tendency for the blood to be shunted from the left ventricle into the aorta than from the right ventricle into the pulmonary artery There is little tendency for a reversal in the direction of the shunt In contrast to most cases of complete transposition of the great vessels the heart does not undergo rapid progressive enlargement

Clinical diagnosis Cyanosis and clubbing are intense Physical development is retarded Stunting of growth may be extreme The heart is but slightly enlarged The shadow at the base of the heart to the left of the sternum is concave and the second sound in this region is accentuated The murmur if present is systolic in time The x ray shows a contour similar to that of a tetralogy of Fallot but the vascular markings extend nearly to the periphery of the lungs The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

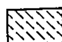
DIAGRAM X-8



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

DIAGRAM X-8

Complete transposition of the great vessels pulmonary stenosis and an auricular septal defect

The essential feature of this malformation is complete transposition of the great vessels combined with pulmonary stenosis the ventricular septum is intact but there is a gross defect in the auricular septum

The blood from the right auricle flows into the right ventricle and is pumped out through the aorta to the body and returned by the superior vena cava and the inferior vena cava to the right auricle In a similar manner the blood from the left auricle flows into the left ventricle and is pumped out through the pulmonary artery to the lungs and returned by the pulmonary veins to the left auricle With the first breaths of life, the pulmonary pressure drops and thus lowers the pressure on the left side of the heart Hence more blood is directed from the right auricle to the left auricle. This continues until the pressure in the left auricle exceeds that of the right auricle thereupon the direction of the shunt is reversed

The pulmonary stenosis protects the lungs but increases the work of the left ventricle As the pressure in the left ventricle rises the pressure in the left auricle is increased The shunt is at the auricular level but it is the relative pressure in the two ventricles which regulates the volume and direction of the shunt.

Clinical diagnosis Cyanosis and clubbing are intense Stunting of growth is extreme The clinical findings and the cardiac findings are similar to those of a complete transposition of the great vessels with pulmonary stenosis and a ventricular septal defect except that there is more strain on the auricles and both the right auricle and left auricle are usually enlarged The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

period of years the extreme polycythemia leads to a decrease in the blood platelets and to the usual changes in the clotting mechanism of the blood

Respirations are rapid, especially in early infancy As the child grows and compensation improves, the respiratory rate slows but always remains accelerated

Barrel shaped chest deformity is the rule

The liver is usually slightly enlarged but does not pulsate

The pulse pressure is narrow The pulses may be so weak that they are difficult to feel, nevertheless, they are of equal strength in the upper and lower extremities

The blood pressure is low

Stunting of growth is usually extreme In no other malformation is the stunting of growth as extreme as that which occurs in transposition of the great vessels A lad of sixteen years may have the stature of a boy of twelve, or a boy of twelve may be as small as the average child of eight Severe stunting of growth in a cyanotic child should always arouse suspicion of a transposition of the great vessels with pulmonary stenosis

Exercise tolerance is extremely limited The child with a complete transposition of the great vessels and pulmonary stenosis seldom learns to walk before three years of age and frequently not until the age of five or six, even then he can walk only a short distance Many of these children get relief by squatting and develop this habit as soon as they learn to walk

CARDIAC FINDINGS

The heart is usually slightly enlarged

A systolic murmur and a *thrill* over the base of the heart to the left of the sternum are the rule Usually, however, neither the murmur nor the thrill is very intense

The second sound at the base is usually better heard to the left of the sternum than to the right, because the abnormal position of the aorta renders the pulmonic second sound louder than the "aortic" second sound There is, however, no reduplication of the second sound

Terminally there may be right sided cardiac failure with engorgement of the liver but congestion in the lungs is minimal

X RAY AND FLUOROSCOPIC FINDINGS

There are two distinct x ray pictures, depending on the position of the aorta

When the aorta arises from the right ventricle but is not rotated abnormally far, the contour of the heart is closely similar to that of a tetralogy of Fallot Nevertheless, during infancy the heart is usually slightly larger and the vascular markings are more conspicuous and extend further toward the periphery of the lungs than in a tetralogy of Fallot

If the pulmonary stenosis is not too severe and the condition is compatible with life for a number of years, as the child grows and the diaphragm descends the contour of the heart comes to resemble that of a tetralogy of Fallot The heart is, however, slightly enlarged There is a concave curve at the base of the heart to the left of the sternum and there are dense hilar shadows In addition there are many small, discrete, circular shadows due to the blood vessels seen on end as they course from the posteriorly placed pulmonary artery to the anterior portion of the lung (see Figure x-27) Careful examination of the x ray film will show



FIGURE X-27 Complete transposition of the great vessels combined with pulmonary stenosis Adult

that the vascular markings extend nearly to the periphery These findings are more readily seen on the x ray film than upon fluoroscopy Hence it is often the discrepancy between these two observations which gives the clue to the diagnosis

In the left anterior oblique position the pulmonary window is clear, as the posteriorly placed pulmonary artery courses to the lungs at a lower level than when it arises from the right ventricle

When the aorta is rotated so far to the left that it occupies the position of the normal pulmonary artery, there is marked fullness of the pulmonary conus, as shown in Figure x–28 The conspicuous size of the 'pulmonary conus' is in striking contrast to the clarity of the lungs In reality the shadow high up to the left of the sternum is cast by the aorta, the branches of the posteriorly placed pulmonary artery are not visible as they course to the lungs In the left anterior oblique position the pulmonary window is large and clear



FIGURE x–28 Complete transposition of the great vessels combined with pulmonary stenosis Child

The aorta is transposed far to the left

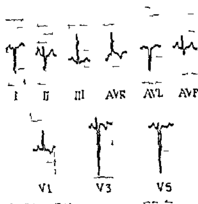


Figure x-29 Complete transposition of the great vessels combined with pulmonary stenosis

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar precordial leads show evidence of right ventricular hypertrophy (see Figure x-29)

SPECIAL TESTS

The condition of an infant with a complete transposition of the great vessels and pulmonary atresia is usually too critical to permit of special studies Hence the following discussion pertains to patients with pulmonary stenosis

The circulation time is abnormally short Two to four seconds from arm to tongue is not unusual

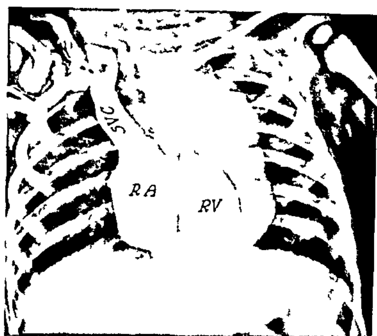
The oxygen saturation of the arterial blood is always reduced and may be extremely low It falls still further with exercise

Cardiac catheterization will usually show that the oxygen content of the blood samples taken from the right auricle, the right ventricle, and the aorta are closely similar It is usually possible to catheterize the aorta but it is manifestly impossible to catheterize the pulmonary artery There may or may not be evidence of an auricular or a ventricular septal defect

Angiocardiography may be of value when the aorta is rotated far to the right, as it will confirm the observation that the vascular shadow at the base of the heart to the left of the sternum is the aorta and not the pulmonary artery (see Figure x-30) Owing to the pulmonary stenosis, no appreciable concentration of dye ever reaches the lungs

DIAGNOSIS

In early infancy the diagnosis is based upon the finding of persistent cyanosis, which appears at an early age and is associated with severe polypnea and fre



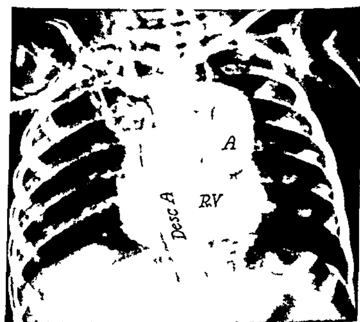


FIGURE X-30 Complete transposition of the great vessels combined with pulmonary stenosis Infant

The aorta rises far to the left and arches to the right

quent attacks of paroxysmal dyspnea, combined with a heart of characteristic contour The concave curve of the cardiac silhouette to the left of the sternum at the base of the heart indicates that the pulmonary artery is diminutive, misplaced, or absent In the left anterior-oblique position the right ventricle is seen to be enlarged The enlargement of the right ventricle and the strength of the pulse indicate that the right ventricle is doing the major work in the maintenance of the circulation The clear pulmonary window indicates that the pulmonary artery is diminutive or absent From these findings it is adduced that the aorta arises mainly from the right ventricle and that the pulmonary artery is markedly stenosed or atretic

In childhood the diagnosis is based upon the finding of a severely incapacitated cyanotic patient with marked stunting of growth who has slight cardiac enlargement and accentuation of the second sound at the base of the heart to the left of the sternum and border line compensation

X ray and fluoroscopic examination aid in the diagnosis The contour of the heart is similar to that of a tetralogy of Fallot but the hilar shadows are dense and the vascular markings extend to the periphery of the lungs When the aorta is rotated far to the left, it occupies the position of the normal pulmonary artery Under such circumstances the aorta arches conspicuously upward to the left of the sternum and the lungs are phenomenally clear

DIFFERENTIAL DIAGNOSIS

The condition is most frequently mistaken for a tetralogy of Fallot, a single ventricle with transposition of the great vessels, or a truncus arteriosus with reduced pulmonary blood flow Occasionally it requires differentiation from a Taussig Bing complex

A tetralogy of Fallot especially one in which there is extreme dextroposition of the aorta, may be extremely difficult to differentiate from a complete transposition of the great vessels with pulmonary stenosis The second sound to the left of the sternum is increased in intensity when the aorta arises from the right ventricle but the same is true when the dextroposition of the aorta is extreme The extension of the vascular markings to the periphery of the lungs is, however, not seen in a tetralogy of Fallot

A single ventricle with pulmonary stenosis may be confused with the malformation under discussion, especially when the great vessels are transposed (see Figure xv–14) Indeed, cardiac catheterization is frequently necessary to differentiate the two conditions

A truncus arteriosus with reduced pulmonary blood flow usually shows a characteristic contour of the heart The heart is large and occupies a transverse position in the chest and the aortic knob is conspicuous Furthermore, the baby, although intensely cyanotic, usually does better than might be anticipated

A Taussig Bing malformation is confused with a complete transposition of the great vessels and pulmonary stenosis only when the aorta arises far to the left In a Taussig Bing malformation there is a full pulmonary conus and the hilar shadows are increased, whereas in the malformation under discussion, the full pulmonary conus is combined with excessively clear lung fields

TREATMENT

When a complete transposition of the great vessels is combined with pulmonary stenosis, the condition may be greatly helped by a systemic pulmonary anastomosis and the creation of an auricular septal defect Although some defect in the auricular or the ventricular septum is essential for life, if only an anastomosis is made, there is great danger that these openings may not be sufficiently large to enable the left side of the heart to expel the increased volume of blood which it receives as a result of the increased circulation to the lungs Therefore, unless a defect of moderate size can be demonstrated, it is wise to combine the anastomotic procedure with the creation of an auricular septal defect Such a defect enables the increased volume of blood which is returned to the left auricle to flow to the right auricle and thence to the right ventricle and out into the aorta From the aorta some blood again flows through the anastomosis to the lungs and is returned to the left auricle Thus the double procedure not only increases the circulation to the lungs but also permits the establishment of a figure of eight circulation Consequently there is hope that this operation may place a constant, not an ever increasing, load upon the heart

A Baffes operation may be of benefit to children with complete transposition of the great vessels and pulmonary stenosis Furthermore, in children it may not be found necessary to relieve the pulmonary stenosis,[14] because the patient can live to childhood only when the pulmonary stenosis is not extreme

PROGNOSIS

Without operation the prognosis is poor Most patients with this malformation are severely incapacitated but may live through childhood Even with operation the prognosis is guarded Although a patient may be symptomatically

benefited by a systemic pulmonary anastomosis combined with the creation of an auricular septal defect, the red blood cell count, the hemoglobin, and the hematocrit values seldom return to normal levels

SUMMARY

Complete transposition of the great vessels may occur in combination with pulmonary atresia but such a malformation is seldom long compatible with life When complete transposition of the great vessels occurs with pulmonary stenosis, the circulation to the lungs is greater but it is still meager, real difficulty is also encountered in the direction of venous blood to the lungs and of oxygenated blood to the systemic circulation

Patients with this malformation are intensely cyanotic Polycythemia devel ops early Dyspnea is marked Stunting of growth is extreme Exercise tolerance is extremely limited These children squat when tired

The heart may be only slightly enlarged A systolic murmur and a thrill are the rule X ray and fluoroscopic findings are closely similar to those of a tetralogy of Fallot with an extreme degree of pulmonary stenosis, except that the vascular shadows are more pronounced and extend to the periphery of the lungs When the aorta is rotated so far to the left that it occupies the position of the main pul monary artery it forms a conspicuous arc above the cardiac shadow Conse quently there is fullness of the pulmonary conus combined with excessively clear lung fields

The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy Cardiac catheterization shows that the aorta arises mainly or entirely from the right ventricle Angiocardiography will demonstrate that the shadow high up to the left of the sternum is the aorta

The malformation requires differentiation from a tetralogy of Fallot with severe pulmonary stenosis, from a single ventricle with pulmonary stenosis, from a truncus arteriosus with reduced pulmonary blood flow, and occasionally from a Taussig Bing malformation

Patients with this malformation may be benefited by a systemic pulmonary anastomosis combined with the creation of an auricular septal defect and also by a Baffes operation

Most of these patients are severely handicapped Without operation the prog nosis is poor Operation improves the circulation but seldom causes a marked reduction in the polycythemia The patient is definitely benefited but still suf fers from some limitation of activity

D *Complete Transposition of the Great Vessels Combined with Complete Interruption of the Aortic Arch*

Complete transposition of the great vessels in combination with complete interruption of the aortic arch presents an unusually distinctive clinical syndrome

NATURE OF THE MALFORMATION

The interruption of the aortic arch usually occurs between the left subclavian artery and the point of entrance of the ductus arteriosus When this occurs, the descending aorta becomes continuous with the pulmonary artery through the ductus arteriosus The heart itself is *normally formed* The ventricular septum is intact (see Figure x-31) The foramen ovale is generally covered by a valve which is not completely sealed

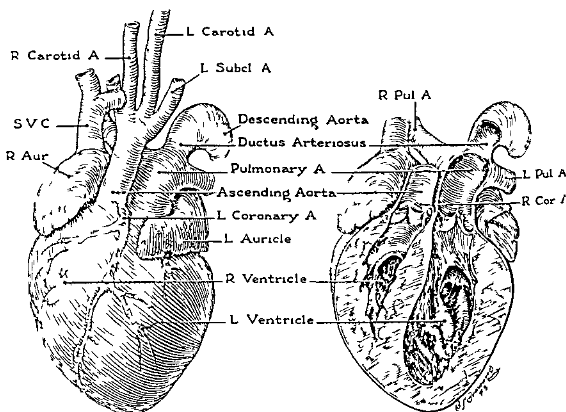


FIGURE x-31 Complete transposition of the great vessels combined with complete interruption of the isthmus of the aorta Infant

The descending aorta is continuous with the pulmonary artery through the ductus arteriosus

COURSE OF THE CIRCULATION

In normal fetal circulation the blood from the pulmonary artery is directed *through* the ductus arteriosus to the descending aorta and some of the blood from the right auricle is directed through the foramen ovale into the left auricle Essentially the same circulatory mechanism persists when there is complete in terruption of the isthmus of the aorta (see Figure xii-2)

When, in addition to the complete interruption of the aortic arch, there is complete transposition of the great vessels, the blood from the right ventricle is pumped out through the aorta to the head and the upper extremities, the blood from the left ventricle is pumped out by way of the pulmonary artery to the lungs and also through the ductus arteriosus to the trunk and the lower ex tremities (see Figure x-32) During fetal life it makes little difference which great vessel pumps the blood to the head and which to the trunk and the lower extremities The malformation places no strain upon the fetal circulation

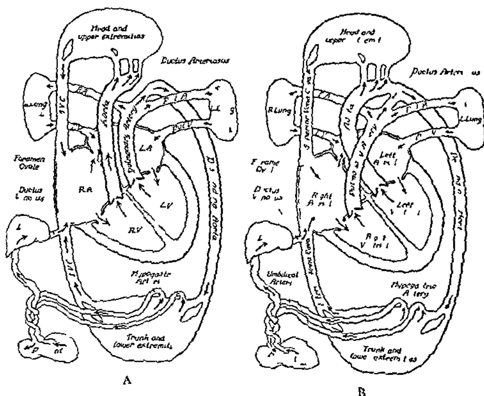


FIGURE x-32 Fetal circulation (A) Complete interruption of the isthmus of the aorta and (B) normal heart

ulation With the expansion of the lungs, the blood in the pulmonary
irected to the lungs The consequence is that the blood supply to the
the lower extremities is meager The blood from the lungs is returned
nal manner to the left auricle With the first breaths of life some blood
cted from the right auricle through the foramen ovale to the left auri
blood from the left auricle flows into the left ventricle and is pumped
gh the pulmonary artery to the lungs As soon as the pressure in the
y artery becomes higher than the pressure in the descending aorta,
d blood will again flow from the pulmonary artery through the ductus
to the descending aorta The blood from the lower extremities is re
the inferior vena cava to the right auricle The blood from the right
ses freely into the right ventricle and is pumped out by way of the
e head and the upper extremities This blood is returned by the su
a cava to the right auricle In addition, the right auricle receives from
r vena cava that portion of the blood from the left ventricle which has
ped through the ductus arteriosus to the descending aorta and the
emities It follows that with each cycle the right auricle receives more
was expelled from the right ventricle during the preceding beat Cor
gly, with each successive beat, the left auricle receives less blood than
umped out from the left ventricle by way of the transposed pulmonary
erefore the pressure in the right auricle will steadily rise and that in
ricle will fall When the difference in the pressure between the right
l the left becomes sufficiently great, the valve which covers the fora
will be forced open and blood will be shunted from the right auricle
This is the only mechanism by which the venous blood is directed to
or oxygenation Diagram x-9 illustrates the course of the circulation

OF THE MALFORMATION

ch as the aorta arises from the right ventricle, the head and the upper
receive venous blood Since the pulmonary artery is continuous with
ding aorta, through the ductus arteriosus, the pressure in the pulmo-
is the same as that in the descending aorta In other words, the lungs
od under systemic pressure Furthermore, the only oxygenated blood
head and the upper extremities ever receive is that which has been
rough the ductus arteriosus to the trunk and the lower extremities
ed by the inferior vena cava to the right auricle This blood is mixed

with the even more poorly oxygenated blood returned to the right auricle from the head and the upper extremities by the superior vena cava Although it is true that normal venous blood is not completely deprived of oxygen, this mixture of blood from the left and the right side of the heart has an extremely meager oxygen content

CLINICAL FINDINGS

Cyanosis of the head and the upper extremities is intense

The difference in cyanosis between the upper and the lower extremities is e most significant of all findings Although the pulses are equal in the upper id the lower extremities, the cyanosis is markedly more intense in the upper xtremities The difference in cyanosis is due to the fact that the lower extremi ies receive oxygenated blood from the descending aorta, which is continuous hrough the patent ductus arteriosus with the pulmonary artery The line of demarcation of the cyanosis lies at the brim of the pelvis The occurrence of the line of demarcation of the cyanosis at this low level indicates that the principal blood supply to the superficial layers of the abdomen is by way of the superficial epigastric arteries the latter arise from the internal mammary arteries, which in turn are branches of the subclavian arteries (see Chapter XXVII) Thus the abdominal wall receives its blood supply from vessels which are given off above the point of interruption of the aorta

Dyspnea and *polypnea* are extreme

The pulse in the lower extremities is of good quality Even though the blood supply is abnormal the strength of the pulse is normal

CARDIAC FINDINGS

The heart enlarges rapidly By four weeks of age it is greatly enlarged A *murmur* may or may not be present If present, it is *systolic* in time Terminally a *gallop rhythm* may develop The liver is enlarged and the lungs are congested

X RAY AND FLUOROSCOPIC FINDINGS

The shape of the heart is significant The shadow at the base of the heart is narrow and the upper margin of the cardiac shadow to the left of the sternum s concave Both ventricles are enlarged The work of the left ventricle is increased because it pumps blood to the lungs as well as to the trunk and the lower extremities whereas the work of the right ventricle is decreased, since it pumps blood only to the head and the upper extremities Consequently the right ventri

DIAGRAM X-9

Head and upper extremities

Ductus Arteriosus

R Lung

L Lung

P.A.

S.V.C.

Aorta

PV

Pulmonary Art.

L.A.

Foramen Ovale

R.A.

L.V.

R.V.

Descending Aorta

I.V.C.

Trunk and lower extremities



Arterial blood (fully saturated)



Small admixture of venous blood
No visible cyanosis



Venous and arterial blood
Cyanosis visible



Venous blood

Diagram X-9

Complete transposition of the great vessels combined with complete interruption of the isthmus of the aorta The descending aorta is continuous with the pulmonary artery through the ductus arteriosus

In this malformation there is in addition to a complete transposition of the great vessels, a complete interruption of the isthmus of the aorta beyond the left subclavian artery, the descending aorta is continuous with the pulmonary artery through the ductus arteriosus Consequently there is always pulmonary hypertension

The blood from the right auricle flows into the right ventricle and is pumped out through the aorta to the head and the upper extremities and is returned by the superior vena cava to the right auricle. The blood in the left auricle flows into the left ventricle and is pumped out into the pulmonary artery The blood which flows to the lungs is returned in the normal manner by the pulmonary veins to the left auricle However some of the blood from the pulmonary artery flows through the ductus arteriosus to the trunk and the lower extremities This blood is returned by the inferior vena cava to the right auricle Thus with each cycle the right auricle receives all the blood pumped out of the right ventricle which is returned by the superior vena cava and some of the blood pumped out of the left ventricle which is returned by the inferior vena cava hence the pressure in the right auricle rises and the pressure in the left auricle falls Consequently the valve covering the foramen ovale is forced open and some blood flows from the right auricle through the foramen ovale to the left auricle This is the only way by which venous blood is directed to the lungs for oxygenation Oxygenated blood is directed to the feet and the lower extremities from the left ventricle through the pulmonary artery by way of the ductus arteriosus The only oxygen which the right side of the heart ever receives is that contained in the venous blood returned by the inferior vena cava Consequently the supply of oxygen to the head and the upper extremities is very meager

Clinical diagnosis The heart is enlarged The shadow at the base of the heart to the left of the sternum is concave The shadow cast by the great vessels is narrow when viewed in the anterior posterior position and increases in width in the left anterior oblique position Cyanosis and dyspnea are intense Furthermore, inasmuch as oxygenated blood is directed from the pulmonary artery through the ductus arteriosus to the descending aorta the lower extremities will be less cyanotic than the upper extremities

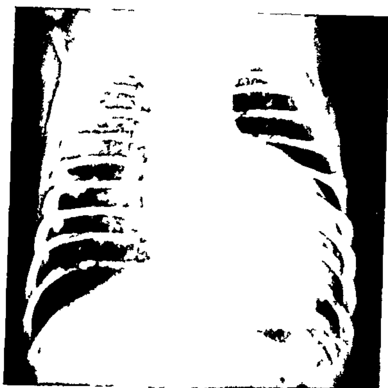


FIGURE X-33 Complete transposition of the great vessels combined with complete interruption of the isthmus of the aorta Infant

cle is not as big as it usually is in most cases of complete transposition of the great vessels and the left ventricle is larger The alteration in the relative size of the two ventricles renders the shape of the heart distinctive The increased size of the left ventricle makes the absence of the pulmonary conus of the right ventricle more conspicuous than it is in most other cases of transposition (compare Figures X-6 and 33) The great size of the left ventricle also causes the apex of the heart to be upturned and the ventricular shadow to extend horizontally toward the axilla In the left anterior oblique position the right ventricle is seen to be enlarged but does not extend as close to the anterior chest wall as in other cases of complete transposition of the great vessels, greater rotation of the infant is required to cause the left ventricle to clear the spinal column

The changes in the width of the shadow at the base of the heart which are characteristic of complete transposition of the great vessels can be demonstrated by fluoroscopic examination In the anterior posterior position the aortic shadow is narrow, upon rotation of the infant into the left anterior oblique position, this shadow increases in width

In addition, rhythmic changes in the size of the right auricle may be seen These are due to the fact that the pressure in the right auricle steadily rises and

that in the left auricle falls until the difference in pressure between the two auricles is sufficient to force open the valve which covers the foramen ovale Thereupon blood will flow from the right auricle to the left auricle and the right auricle will collapse

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads usually show a right axis deviation and the unipolar precordial leads should show evidence of greater hypertrophy of the left ventricle than occurs in other cases of complete transposition of the great vessels

DIAGNOSIS

The diagnosis is based upon the finding of an infant with intense cyanosis and severe dyspnea whose feet and lower extremities are of almost normal color The heart is greatly enlarged In the anterior posterior x ray film there is absence of the fullness of the pulmonary conus, a sharp concave curve to the left of the sternum, and a narrow aortic shadow In the left anterior-oblique position the shadow at the base of the heart is increased in width and both ventricles are seen to be enormously enlarged, the right ventricle, however, is slightly smaller and the left ventricle is even larger than is usual in a complete transposition of the great vessels

DIFFERENTIAL DIAGNOSIS

The condition is to be differentiated (1) from a complete transposition of the great vessels with a patent ductus arteriosus the foramen ovale covered by a valve, and a normal aortic arch and (2) from complete interruption of the isthmus of the aorta without complete transposition of the great vessels

Complete transposition of the great vessels with a patent ductus arteriosus and the foramen ovale covered by a valve which is not completely sealed is physiologically closely similar to the malformation under discussion The contours of the heart in the anterior posterior position are identical (compare Figures x–10 and 33) The two conditions can be differentiated by the difference in the intensity of the cyanosis between the upper and the lower extremities in the malformation under discussion the difference is readily apparent, whereas when the aorta is intact it may be so slight that it is difficult to determine with certainty

Complete interruption of the isthmus of the aorta without a transposition of the great vessels can readily be differentiated from the malformation under discussion by the distribution of the cyanosis When the great vessels occupy their

normal position, only the feet and the lower extremities are cyanotic, whereas when the great vessels are transposed, the cyanosis is limited to the head, the upper extremities, and the abdominal wall In both instances the line of demarcation of the cyanosis lies at the brim of the pelvis When the cyanosis is limited to the feet, it may be completely overlooked or thought to be due to cold Furthermore, dyspnea is absent

PROGNOSIS

The prognosis is extremely poor The oxygen supply to the head and the upper extremities is so meager that the malformation is compatible with life for only a few weeks

SUMMARY

The diagnostic features of this malformation are (1) intense cyanosis and severe dyspnea, (2) the features which are pathognomonic of a complete transposition of the great vessels *the narrow aortic shadow in the anterior* posterior position and the wide aortic shadow in the left anterior-oblique position, combined with enlargement of both ventricles and the absence of the fullness of the normal pulmonary conus of the right ventricle, (3) the features which are characteristic of complete absence of the aortic arch between the left subclavian artery and the point of entrance of the ductus arteriosus although the pulses are of equal strength in both extremities, there is a difference in the intensity of cyanosis between the upper and lower extremities, when this occurs in combination with a complete transposition of the great vessels, the cyanosis of the upper extremities is more intense than that of the lower extremities, (4) rhythmic changes in the size of the right auricle indicative of a foramen ovale which is at times functionally open and at times functionally closed

References

1 Rokitansky, C von Die Defekte der Scheidewande der Herzens Vienna Braumuller, 1875
2 Spitzer, A Ueber den Bauplan des normalen und missbildeten Herzens Versuch einer phylogenetischen Theorie Virchows Arch f path Anat 243 81–272 1923
3 Spitzer, A The architecture of normal and malformed hearts a phylogenetic theory of their development, with a summary and analysis of the theory by Maurice Lev and Aloysius Vass Springfield Ill Thomas, 1951
4 Pernkopf, E and W Wirtinger Die Transposition der Herzostien ein Versuch der

Erklarung dieser Erscheinung Ztschr f Anat u Entwcklngsgesch 100 563-711, 1933

5 Harris J S and S Farber Transposition of the great cardiac vessels with special reference to the phylogenetic theory of Spitzer Arch Path 28 427-502 1939

6 Bremer J L Transposition of the aorta and the pulmonary artery an embryologic study of its cause Arch Path 34 1016-1030 1942

7 Emerson P W and H Green Transposition of the great cardiac vessels J Pediat 21 1-22, 1942

8 Lev, M and O Saphir A theory of transposition of arterial trunks based on the phylogenetic and ontogenetic development of the heart Arch Path 39 172-183, 1945

9 Edwards J Personal communication

10 Taussig H B Complete transposition of the great vessels clinical and pathologic features Am Heart J 16 7 8-733 1938

11 Mannheimer E Personal communication

12 Blalock A and C R Hanlon The surgical treatment of complete transposition of the aorta and the pulmonary artery Surg Gynec & Obst 90 1-15 1950

13 Baffes T G A new method for surgical correction of transposition of the aorta and pulmonary artery Surg., Gynec & Obst 102 227-233 1956

14 Miller R. Personal communication

CHAPTER XI

THE TAUSSIG-BING MALFORMATION

A TRANSPOSITION of the aorta combined with a pulmonary arter which arises from the right ventricle and partially over rides the ven tricular septum was listed by Pernkopf in 1926[1] as one possible com bination of anomalies in a transposition of the great vessels It was not, howeve until 1948 that Taussig and Bing[2] showed that this malformation presented distinctive clinical syndrome

This malformation is relatively common among children who suffer fron persistent cyanosis because the structure of the heart permits a fair degree of th crossing of the two circulations and does not lead to progressive cardiac enlarge ment Therefore the condition is compatible with relative longevity Failure t differentiate this syndrome as a specific entity occurred partly because the anom aly is compatible with relative longevity and partly because both clinicians an pathologists confused it with an Eisenmenger complex The two conditions are however, quite different In the Eisenmenger complex the aorta arises mainl from the left ventricle and partially over rides the ventricular septum and con sequently it receives blood from both ventricles, the pulmonary artery arises en tirely from the right ventricle In contrast to this, in the Taussig Bing malforma tion the aorta is transposed and arises entirely from the right ventricle, the pulmonary artery also arises from the right ventricle but partially over rides the ventricular septum, consequently it is the pulmonary artery that receives blooc from both ventricles

The Taussig Bing malformation is also closely similar to the condition in which both the aorta and the pulmonary artery arise from the right ventricle (see Chapter vii) In such an anomaly there is always a ventricular septal defect to enable the blood to escape from the left ventricle Abbott[3] warned that this latter malformation should not be confused with an Eisenmenger complex The defect frequently lies at the base of the pulmonary artery, which, however, does not over ride the opening When the pulmonary artery over rides the ventricular septum the condition is more readily compatible with life than when both great vessels arise from the right ventricle Consequently such is the usual nature of the malformation among those who live until childhood or early adult life

NATURE OF THE MALFORMATION

The outstanding features of this malformation are the aorta is transposed and arises entirely from the right ventricle, the pulmonary artery arises mainly from the right ventricle but partially over rides the ventricular septum and consequently also receives blood from the left ventricle, there is a high ventricular septal defect, and there is right ventricular hypertrophy

In this malformation the transposed aorta lies to the right of the pulmonary artery, as shown in Figure XI-1 The aorta is usually smaller than the pulmonary artery the aorta, however, may be normal in size and the pulmonary artery is frequently abnormally large When the pulmonary artery over rides the ventricular septum, the condition is described as levoposition of the pulmonary artery to designate the counterpart of dextroposition of the aorta *Dextroposition of the* aorta means that, although the aorta arises primarily from the left ventricle, it is displaced so that the aortic orifice partially over rides the ventricular septum and consequently *the aorta receives blood directly from the right ventricle* Levo-

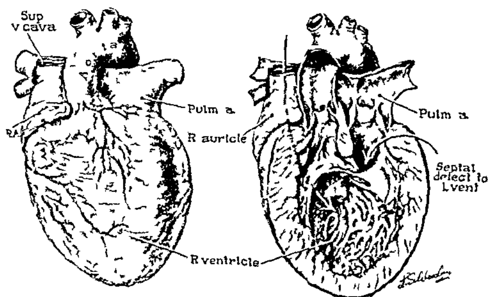


FIGURE XI-1 Taussig Bing malformation (Case XI-1 same patient as in Figures XI-4 5 6) Child

CHAPTER XI

THE TAUSSIG-BING MALFORMATION

A TRANSPOSITION of the aorta combined with a pulmonary artery which arises from the right ventricle and partially over-rides the ventricular septum was listed by Pernkopf in 1926[1] as one possible combination of anomalies in a transposition of the great vessels. It was not, however, until 1948 that Taussig and Bing[2] showed that this malformation presented a distinctive clinical syndrome.

This malformation is relatively common among children who suffer from persistent cyanosis because the structure of the heart permits a fair degree of the crossing of the two circulations and does not lead to progressive cardiac enlargement. Therefore the condition is compatible with relative longevity. Failure to differentiate this syndrome as a specific entity occurred partly because the anomaly is compatible with relative longevity and partly because both clinicians and pathologists confused it with an Eisenmenger complex. The two conditions are, however, quite different. In the Eisenmenger complex the aorta arises mainly from the left ventricle and partially over-rides the ventricular septum and consequently it receives blood from both ventricles; the pulmonary artery arises entirely from the right ventricle. In contrast to this, in the Taussig-Bing malformation the aorta is transposed and arises entirely from the right ventricle; the pulmonary artery also arises from the right ventricle but partially over-rides the ventricular septum; consequently it is the pulmonary artery that receives blood from both ventricles.

The Taussig-Bing malformation is also closely similar to the condition in which both the aorta and the pulmonary artery arise from the right ventricle (see Chapter VII). In such an anomaly there is always a ventricular septal defect to enable the blood to escape from the left ventricle. Abbott[3] warned that this latter malformation should not be confused with an Eisenmenger complex. The defect frequently lies at the base of the pulmonary artery, which, however, does not over-ride the opening. When the pulmonary artery over-rides the ventricular septum the condition is more readily compatible with life than when both great vessels arise from the right ventricle. Consequently such is the usual nature of the malformation among those who live until childhood or early adult life.

ventricle is also pumped into the pulmonary artery (see Figure XI-3) Thus the pulmonary artery receives blood from both ventricles Consequently the volume of blood which flows through the pulmonary artery during fetal life is greater than normal This may be the explanation of the abnormally large size of the pulmonary artery

After birth all the blood from the right auricle flows into the right ventricle Part of the blood from the right ventricle is pumped out by way of the aorta to the systemic circulation and part is pumped out by way of the pulmonary artery to the lungs All the blood which goes to the lungs is returned by the pulmonary veins to the left auricle Thence it flows into the left ventricle Most of the blood from the left ventricle is pumped directly into the pulmonary artery, which over rides the ventricular septum and arises in part from the left ventricle This oxy genated blood re-circulates through the lungs and is returned to the left auricle and the left ventricle Inasmuch as the aorta arises from the right ventricle, the only oxygenated blood which reaches the aorta is that which is pumped through

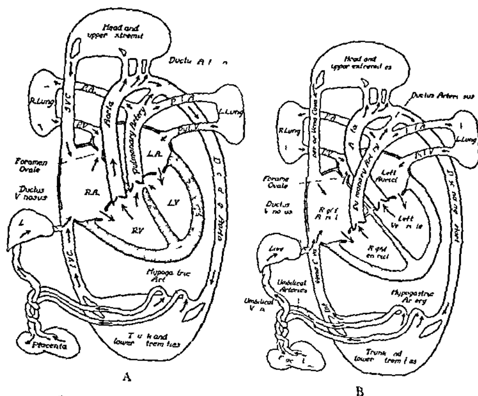


FIGURE XI-3 Fetal circulation (A) Taussig Bing malformation and (B) normal heart

position of the pulmonary artery means that the pulmonary artery, although it arises mainly from the right ventricle, partially over rides the ventricular septum and thereby receives blood from both ventricles, as shown in the cross section in Figure xi-2 A high ventricular septal defect is inevitable either with dextroposition of the aorta or with levoposition of the pulmonary artery In the malformation under discussion the abnormal position of the pulmonary artery causes the defect to lie beneath that vessel Inasmuch as the aorta arises entirely from the right ventricle and the pulmonary artery also arises mainly from the right ventricle, there is right ventricular hypertrophy

COURSE OF THE CIRCULATION

During fetal life the right ventricle does the main work Most of the blood from the right auricle flows into the right ventricle and from there is pumped out both into the aorta and into the pulmonary artery The blood in the aorta is directed to the systemic circulation and is returned in the normal manner to the right auricle The blood in the pulmonary artery flows to the lungs and through the ductus arteriosus to the descending aorta The blood which flows to the trunk and the lower extremities is returned in the normal manner to the right auricle and that which circulates through the lungs is returned in the normal fashion to the left auricle and thence to the left ventricle Inasmuch as the aorta arises entirely from the right ventricle and the pulmonary artery arises partially from the left ventricle, the blood from the left ventricle is pumped out into the pulmonary artery As previously mentioned, part of the blood from the right

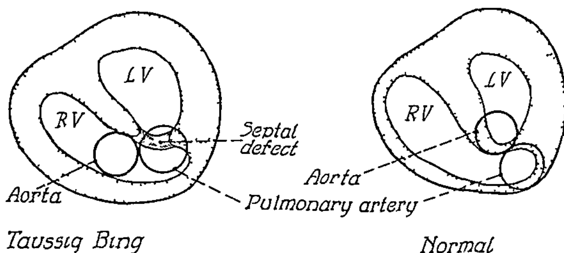


FIGURE xi-2 Taussig Bing malformation and normal heart

between the gradual expansion of the lungs and the pulmonary vascular changes Eventually the pulmonary resistance becomes so high that the right-to-left shunt is increased and the oxygen saturation of the arterial blood is further decreased The collateral circulation develops, polycythemia increases still further and may become extreme

Polypnea is a common complaint Infants with this malformation rarely have attacks of paroxysmal dyspnea such as so frequently occur in infants with pulmonary stenosis or atresia, and they do not suffer from episodes of loss of consciousness such as are common in infants with a complete transposition of both great vessels

The exercise tolerance is limited Nevertheless, the incapacity is not as great as might be expected with a transposed aorta Although the child may tend to *squat when he first starts to walk*, he soon overcomes the habit He can frequently walk a considerable distance at a slow pace

Stunting of growth may occur *Weight gain* is slow and often the child is very thin

The blood pressure is difficult to obtain and *the pulse pressure* is narrow but of equal strength in the upper and the lower extremities

CARDIAC FINDINGS

The heart is but slightly enlarged It may or may not be possible to detect the fullness of the pulmonary conus by percussion The *second sound* over the pulmonary area is accentuated On palpation there is a *systolic thrill* and on auscultation a *systolic murmur* is audible over the precordium The murmur is usually neither very loud nor rasping

X-RAY AND FLUOROSCOPIC FINDINGS

The heart is virtually of normal size The pulmonary conus is full and the hilar markings are increased (see Figure XI-4) In early childhood the contour of the heart may closely resemble that of an uncomplicated patent ductus arteriosus or an auricular septal defect In older children and young adults the contour of the heart is closely similar to that of an Eisenmenger complex

The left anterior-oblique position is the most advantageous for the study of the aorta and the pulmonary artery In this view the aorta appears small and the large pulmonary artery arches posteriorly below the aorta

The right anterior-oblique position shows the relation of the pulmonary artery to the esophagus When the pulmonary artery is abnormally large, it may

the septal defect into the right ventricle In the right ventricle the oxygenated blood from the left ventricle mixes with the venous blood from the right auricle, this mixture of oxygenated and venous blood is pumped out by way of the aorta to the systemic circulation and is returned in the normal manner by the superior and inferior venae cavae to the right auricle There the cycle starts again The course of the circulation is shown in Diagram XI-1

PHYSIOLOGY OF THE MALFORMATION

Inasmuch as the aorta arises from the right ventricle, the right ventricle takes over the function of the left ventricle, the pressure in the right ventricle is that which is normal for the left ventricle Consequently the pulmonary artery, which arises from the right ventricle, receives blood under systemic pressure, hence there is always pulmonary hypertension

The pressure in the left ventricle is also elevated, as at best it must expel blood against systemic pressure, in addition, there is difficulty in the ejection of the large volume of blood returned to the left ventricle, for this reason the pressure in the left ventricle may exceed that in the right ventricle This in turn further raises the pressure in the pulmonary artery Consequently the pressure in the pulmonary artery may exceed the systemic pressure Since the oxygenated blood returned to the left ventricle is directed into the pulmonary artery, the oxygen saturation of the blood in the pulmonary artery is higher than that in the aorta

The high pressure with which the blood is ejected into the lungs sets up the usual series of changes in the pulmonary vascular bed As the pulmonary resistance rises, less blood reaches the lungs for oxygenation and more blood is shunted into the aorta As the years go by, these changes gradually lead to a further decrease in the oxygen saturation of the arterial blood and to ever increasing polycythemia Eventually the condition becomes incompatible with life

CLINICAL FINDINGS

The outstanding clinical findings are persistent cyanosis and clubbing of the extremities

Cyanosis dates from birth and is of uniform distribution *Clubbing* of the extremities develops at an early age There is usually dilatation of all the capillaries of the body and suffusion of the conjunctivae

Polycythemia is the rule The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are abnormally high During childhood the hemoconcentration remains relatively constant, as there is a balance

DIAGRAM XI-I

Taussig Bing malformation

In this malformation the aorta is transposed and arises from the right ventricle The pulmonary artery arises mainly from the right ventricle but slightly over rides the ven tricular septum and receives blood *from both ventricles* It follows that the ventricular septal defect is an integral part of the malformation The defect lies beneath the pul monary artery Since the aorta arises from the right ventricle and the pulmonary artery also arises in part from the right ventricle there is pulmonary hypertension

The blood from the right auricle *flows into the right ventricle* Inasmuch as the aorta arises from the right ventricle part of the blood from the right ventricle is pumped directly into the aorta and part is pumped into the pulmonary artery The blood pumped into the pulmonary artery flows to the lungs, where it is oxygenated and is returned to the left auricle in the normal manner All the blood in the left auri cle flows into the left ventricle The blood in the left ventricle is pumped out into the pulmonary artery and through the septal defect into the right ventricle Inasmuch as the pulmonary artery over rides the ventricular septum it receives a mixture of oxy genated blood from the left ventricle and venous blood from the right ventricle. This mixture of oxygenated and venous blood circulates through the lungs and is returned to the left side of the heart The only oxygenated blood which the aorta receives is that which is pumped from the left ventricle through the septal defect into the right ventri cle. Thus mixture of oxygenated and venous blood which has a lower oxygen content than that in the pulmonary artery is pumped into the aorta and circulates through the body and is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

Clinical diagnosis Cyanosis dates from birth and is persistent Polycythemia and clubbing of the extremities develop at an early age The patient s exercise tolerance is limited but he does not squat when tired The heart is but slightly enlarged The con tour of the heart is similar to that seen in an Eisenmenger complex in that there is fullness of the pulmonary conus Pulsations in the hilar shadows develop at an early age The fact that both the aorta and the pulmonary artery arise from the right ventri cle increases the work of that chamber The electrocardiogram shows a right axis devi ation and evidence of right ventricular hypertrophy

DIAGRAM XI-1

Head and upper extremities

R Lung

L Lung

S.V.C.

P.A.

P.V.

R.P.

Aorta

Pulmonary Artery

PV

L A

R.A.

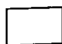
L V

R.V.

Descending Aorta

I.V.C.

Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

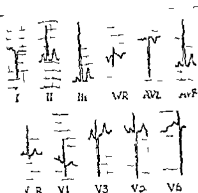


FIGURE XI-5 Taussig Bing malformation (Case XI-1) Child

into the right diverts the catheter from the pulmonary orifice. If the catheter does not enter the pulmonary artery, the findings are similar to those in a tetralogy of Fallot. If the pulmonary artery is catheterized, the pressure in the pulmonary artery is as high or higher than that in the aorta and the oxygen content of the blood in the pulmonary artery is higher than that in the aorta.

Angiocardiography reveals early filling of the aorta and slight opacification in the region of the pulmonary conus. The dye may be dissipated so rapidly through the large pulmonary artery that little of the radio-opaque material is visible in the lungs (see Figure XI-6).

The procedure, however, is not without danger because the rapid injection of the dye increases the pressure in the right side of the heart and consequently may block the flow of blood from the left ventricle to the right ventricle and thereby may decrease the supply of oxygen to the body. Consequently, not only is a large amount of contrast media poured into the aorta, but the patient is simultaneously deprived of oxygen. The procedure has been known to cause sudden death. Usually, however, the oxygen saturation of the arterial blood is sufficiently high so that the momentary decrease in the oxygen content of the blood is not dangerous for the patient. Nevertheless caution should be exercised, especially as angiocardiography is seldom necessary for the diagnosis of this malformation.

DIAGNOSIS

The diagnosis is based upon the contour of the heart in the x ray and the finding of persistent cyanosis which dates from birth. The heart is but slightly enlarged and the x ray shows fullness of the pulmonary conus and increased hilar shadows. Fluoroscopy usually reveals expansile pulsations in the hilar vessels. The electrocardiogram shows a right axis deviation and evidence of right ven



FIGURE XI-4 Taussig Bing malformation
(Case XI-1) Child

cause backward displacement of the esophagus below the level of the aortic arch

Fluoroscopic examination usually reveals expansile pulsations of the hilar shadows, these become increasingly conspicuous with age

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar precordial leads show evidence of right ventricular hypertrophy (see Figure XI-5)

SPECIAL TESTS

The circulation time (arm to tongue) is abnormally short

The oxygen saturation of the arterial blood is abnormally low Upon exercise there is a gradual further fall in the oxygen content of the arterial blood

Cardiac catheterization reveals a high pressure in the right ventricle The oxygen content of the blood in the right ventricle is higher than that in the right auricle It is easy to pass the catheter into the aorta In some instances it is possible to catheterize the aorta and the pulmonary artery with equal ease In other instances difficulty may be encountered in catheterization of the pulmonary artery because the stream of blood which is pumped from the left ventricle

tricular hypertrophy The diagnosis is confirmed by catheterization when the aorta and the pulmonary artery are entered with equal ease

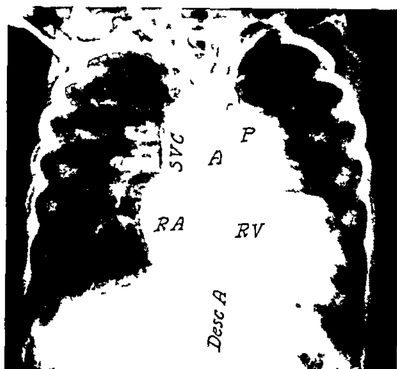
DIFFERENTIAL DIAGNOSIS

The malformation is most commonly confused with an Eisenmenger complex and occasionally with a complete transposition of the great vessels and a dilated pulmonary artery, it may also be confused with a cor pulmonale, that is, with primary pulmonary hypertension

The Eisenmenger complex produces a clinical syndrome which, in older patients, is closely similar to that of the Taussig Bing malformation In both malformations the patients show persistent cyanosis, in neither does the patient squat The contours of the heart and the x ray and fluoroscopic findings are similar both malformations cause fullness of the pulmonary conus and a hilar dance Both show a right axis deviation and evidence of right ventricular hypertrophy Both have a short circulation time The salient difference between the two malformations is that in the Taussig Bing malformation cyanosis dates from birth, whereas in the Eisenmenger complex cyanosis usually develops at or about the time of puberty Hence, if the clinical picture is that of an Eisenmenger complex except that cyanosis dates from birth, the malformation is probably that of the Taussig Bing type

Complete transposition of the great vessels with a dilated pulmonary artery which over rides the ventricular septum is functionally similar to a Taussig Bing malformation [4] When the great vessels are completely transposed, the occurrence of a concave curve at the base of the heart and large blotchy hilar shadows clearly differentiates it from a Taussig Bing malformation Occasionally the main pulmonary artery is so greatly dilated that it is visible to the left of the sternum and gives the appearance of a normally placed dilated pulmonary artery Under such circumstances a complete transposition of the great vessels may *closely* simulate a Taussig Bing malformation Angiocardiography or observation of the course of the catheter as it enters the pulmonary artery may be necessary to differentiate the two conditions

A partial anomaly of the venous return combined with a Taussig Bing malformation has been reported by Schultz [5] Such an anomaly is of benefit to the patient, as the entrance of some of the pulmonary veins into the right auricle raises the oxygen saturation of the blood in the right auricle and in the right ventricle and increases the volume of oxygenated blood directed into the aorta (see Diagram XI–2) Hence it lessens the patient s incapacity and renders the



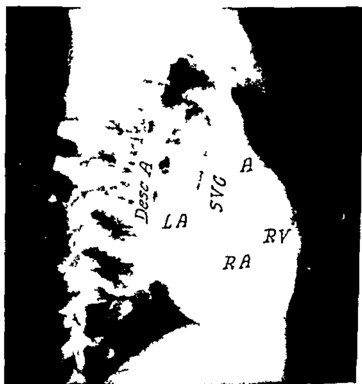


FIGURE XI–6 Taussig Bing malformation (Case XI–1) Child

Anterior posterior view shows simultaneous opacification of the aorta and the pulmonary artery In the lateral view the pulmonary artery is concealed by the aorta

tricular hypertrophy The diagnosis is confirmed by catheterization when the aorta and the pulmonary artery are entered with equal ease

DIFFERENTIAL DIAGNOSIS

The malformation is most commonly confused with an Eisenmenger complex and occasionally with a complete transposition of the great vessels and a dilated pulmonary artery, it may also be confused with a cor pulmonale, that is, with primary pulmonary hypertension

The Eisenmenger complex produces a clinical syndrome which, in older patients, is closely similar to that of the Taussig Bing malformation In both malformations the patients show persistent cyanosis, in neither does the patient squat The contours of the heart and the x ray and fluoroscopic findings are similar both malformations cause fullness of the pulmonary conus and a hilar dance Both show a right axis deviation and evidence of right ventricular hypertrophy Both have a short circulation time The salient difference between the two malformations is that in the Taussig Bing malformation cyanosis dates from birth whereas in the Eisenmenger complex cyanosis usually develops at or about the time of puberty Hence, if the clinical picture is that of an Eisenmenger complex except that cyanosis dates from birth, the malformation is probably that of the Taussig Bing type

Complete transposition of the great vessels with a dilated pulmonary artery which over rides the ventricular septum is functionally similar to a Taussig Bing malformation [4] When the great vessels are completely transposed the occurrence of a concave curve at the base of the heart and large blotchy hilar shadows clearly differentiates it from a Taussig Bing malformation Occasionally the main pulmonary artery is so greatly dilated that it is visible to the left of the sternum and gives the appearance of a normally placed dilated pulmonary artery Under such circumstances a complete transposition of the great vessels may closely simulate a Taussig Bing malformation Angiocardiography or observation of the course of the catheter as it enters the pulmonary artery may be necessary to differentiate the two conditions

A partial anomaly of the venous return combined with a Taussig Bing malformation has been reported by Schultz [5] Such an anomaly is of benefit to the patient as the entrance of some of the pulmonary veins into the right auricle raises the oxygen saturation of the blood in the right auricle and in the right ventricle and increases the volume of oxygenated blood directed into the aorta (see Diagram XI-2) Hence it lessens the patient s incapacity and renders the

DIAGRAM XI-2

Head and upper extremities

R Lung

S.V.C.

P.A.

Aorta

Pulmonary Artery

P.A.

L Lung

PV

PV

L A

Descending Aorta

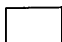
R.A.

L V

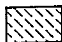
R V

I.V.C.

Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis

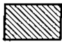

Venous blood

DIAGRAM XI-2

Taussig Bing malformation combined with a partial anomaly of the pulmonary venous return

In this instance a Taussig Bing malformation is combined with the anomalous return of the right pulmonary veins into the right auricle

The blood from the right auricle flows into the right ventricle Since the aorta arises entirely from the right ventricle and the pulmonary artery arises partly from the right ventricle the blood from the right ventricle is pumped out into both the aorta and the pulmonary artery hence there is pulmonary hypertension The blood which goes to the lungs is oxygenated Part of the oxygenated blood from the lungs is returned to the right auricle and the rest is returned to the left auricle thence it flows to the left ventricle The blood in the left ventricle is pumped out into the pulmonary artery and through the septal defect into the right ventricle and thence into the aorta All the blood which is directed into the aorta is returned by the superior vena cava and the inferior vena cava to the right auricle There the venous blood is mixed with the fully oxygenated blood which is returned by the right pulmonary veins to the right auricle This mixture of venous and arterial blood flows into the right ventricle. There the cycle starts again

Clinical diagnosis The diagnosis is based upon the finding of a slightly cyanotic patient with the cardiac findings of a Taussig Bing malformation Cyanosis is minimal because part of the blood which is oxygenated in the lungs is returned by the anomalous vein to the right auricle and thence to the right ventricle thereby the volume of the oxygenated blood directed to the systemic circulation is increased

ondition more compatible with life This combination of anomalies should be onsidered when the physical findings are similar to those in a Taussig Bing nalformation but cyanosis is minimal

A gross defect in the auricular septum is also beneficial to the patient Indeed, uch a defect does what the surgeon attempts to do when he creates an auricular efect (see below)

Cor pulmonale and primary pulmonary hypertension in older patients may esemble the Taussig Bing malformation However, a patient with a cor pul ionale or primary pulmonary hypertension does not usually show cyanosis at irth Indeed, the development of cyanosis during adolescence or in early adult fe is characteristic of a cor pulmonale Moreover, the severity of the dyspnea is ut of proportion to the intensity of the cyanosis Furthermore, the systemic irculation time is normal or prolonged in patients with primary pulmonary hy ertension, whereas in the malformation under discussion the arm to-tongue cir ulation time is abnormally short Either cardiac catheterization or angiocardi graphy will clearly differentiate the two malformations, as the aorta arises from ie left ventricle in a cor pulmonale and from the right ventricle in a Taussig ing malformation

REATMENT

Surgery offers some hope to patients with a Taussig Bing malformation The reation of an auricular septal defect aids in the direction of oxygenated blood o the systemic circulation, and thereby lessens the incapacity of the patient, but does nothing to alter the pulmonary hypertension

A Baffes operation[6] is also of real benefit to a patient with a Taussig Bing nalformation, as it aids in the direction of oxygenated blood to the aorta and f venous blood to the pulmonary artery Neither the Baffes operation nor the reation of an auricular septal defect reduces the pressure in the pulmonary ar ery Unfortunately the pulmonary hypertension will eventually cause difficulty

Medical treatment is the same as for other cyanotic children Intravenous uid should always be given extremely slowly, because any procedure which in reases the pressure in the right auricle and the right ventricle will lessen the olume of oxygenated blood which is shunted from the left ventricle to the right entricle and consequently it will reduce the supply of oxygen to the body

ROGNOSIS

The prognosis varies with the size of the aorta, with the size and position of ie pulmonary artery, and with the size of the septal defect Unless the aorta is

markedly hypoplastic or the septal defect is so small that little oxygenated blood can reach the aorta, the patient generally lives to adult life Nevertheless, the long-continued pulmonary hypertension leads to secondary changes in the lungs, which permit less blood to flow through the pulmonary artery to the lungs, and hence a greater volume of venous blood is directed into the aorta The condition eventually becomes incompatible with life

SUMMARY

The anatomical features of this malformation are a transposed aorta, a large pulmonary artery which arises from the right ventricle and slightly over rides the ventricular septum, a high ventricular septal defect, and right ventricular hypertrophy

The outstanding clinical features are cyanosis which dates from birth, polycythemia, and clubbing of the extremities, combined with a heart which is not greatly enlarged The x ray contour of the heart is similar to that of an Eisenmenger complex because of the fullness of the pulmonary conus, the increased hilar shadows, and the fluoroscopic evidence of a hilar dance The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy The oxygen saturation of the arterial blood is abnormally low The circulation time is abnormally short

Cardiac catheterization shows a high pressure in the right ventricle and the oxygen content of the blood in the right ventricle is higher than that in the right auricle It may be possible to catheterize both the aorta and the pulmonary artery with equal ease if so the diagnosis is readily established by the finding that both the oxygen content of the blood in the pulmonary artery and the pressure in the pulmonary artery are as high or higher than those in the aorta

Angiocardiography, although not usually necessary for diagnosis, may be of value in the rare case in which the contour of the heart in a Taussig Bing malformation simulates that of a complete transposition of the great vessels with a dilated pulmonary artery

The diagnosis is based upon the demonstration of a heart with a contour and fluoroscopic findings similar to those of an Eisenmenger complex in a patient with a history of cyanosis which dates from birth The diagnosis is substantiated by cardiac catheterization provided both the aorta and the pulmonary artery are catheterized and pressures are measured The patient s exercise tolerance is limited nevertheless, relative longevity is the rule

If the patient is extremely incapacitated, the condition may be ameliorated by the creation of an auricular septal defect or by a Baffes operation Such opera

condition more compatible with life This combination of anomalies should be considered when the physical findings are similar to those in a Taussig Bing malformation but cyanosis is minimal

A gross defect in the auricular septum is also beneficial to the patient Indeed, such a defect does what the surgeon attempts to do when he creates an auricular defect (see below)

Cor pulmonale and primary pulmonary hypertension in older patients may resemble the Taussig Bing malformation However, a patient with a cor pulmonale or primary pulmonary hypertension does not usually show cyanosis at birth Indeed, the development of cyanosis during adolescence or in early adult life is characteristic of a cor pulmonale Moreover, the severity of the dyspnea is out of proportion to the intensity of the cyanosis Furthermore, the systemic circulation time is normal or prolonged in patients with primary pulmonary hypertension, whereas in the malformation under discussion the arm to-tongue circulation time is abnormally short Either cardiac catheterization or angiocardiography will clearly differentiate the two malformations, as the aorta arises from the left ventricle in a cor pulmonale and from the right ventricle in a Taussig Bing malformation

TREATMENT

Surgery offers some hope to patients with a Taussig Bing malformation The creation of an auricular septal defect aids in the direction of oxygenated blood to the systemic circulation, and thereby lessens the incapacity of the patient, but it does nothing to alter the pulmonary hypertension

A Baffes operation[6] is also of real benefit to a patient with a Taussig Bing malformation, as it aids in the direction of oxygenated blood to the aorta and of venous blood to the pulmonary artery Neither the Baffes operation nor the creation of an auricular septal defect reduces the pressure in the pulmonary artery Unfortunately the pulmonary hypertension will eventually cause difficulty

Medical treatment is the same as for other cyanotic children Intravenous fluid should always be given extremely slowly because any procedure which increases the pressure in the right auricle and the right ventricle will lessen the volume of oxygenated blood which is shunted from the left ventricle to the right ventricle and consequently it will reduce the supply of oxygen to the body

PROGNOSIS

The prognosis varies with the size of the aorta, with the size and position of the pulmonary artery, and with the size of the septal defect Unless the aorta is

per cent The oxygen capacity was 30.8 volumes per cent The pressure in the right ventricle was 42/19 mm of mercury and that in the pulmonary artery was 57/45 mm of mercury

Angiocardiography was performed by the technique customary in 1947 This necessitated two injections in order to obtain films in two planes Unfortunately there was a mechanical failure at the time of the second injection and therefore a third injection* was given fifteen minutes later Three minutes thereafter the child sat bolt upright and the heart stopped All efforts at resuscitation failed

The angiocardiogram showed that the dye entered the right auricle and then the right ventricle, immediately thereafter the aorta was promptly opacified Little dye was seen in the pulmonary artery or the lungs In the final series of films taken in the lateral position the aorta appeared to arise from the anterior portion of the right ventricle but in this series, too the dye could not be traced into the lungs or to the left side of the heart (see Figure XI-6)

Final clinical diagnosis A transposition of the great vessels was indicated by the physiological studies and angiocardiograms x ray and fluoroscopic studies, however, indicated that the pulmonary artery arose in its normal position

Autopsy No 31039 (performed by Dr Edmund Novak) The chief interest centered about the heart It weighed 180 gm The right auricle was not greatly enlarged The superior vena cava and the inferior vena cava opened into it in the normal fashion The foramen ovale was completely covered by a valve but the margin of the valve showed a probe patency for a distance of 1.0 cm The tricuspid valve which was slightly thickened opened into the right ventricle That chamber was tremendously hypertrophied its wall measured 1 5 cm in thickness The pulmonary artery arose approximately in its normal position The aorta was transposed, it arose entirely from the right ventricle The aortic orifice lay adjacent to both the pulmonary orifice and the ventricular septum as shown in Figures XI-1 and 2 The aortic valve had three cusps and the coronary arteries were given off from the aorta in the normal manner The aortic ring measured 3 5 cm in circumference The aorta and its branches appeared to be normal The maximum circumference of the ascending aorta was 4.0 cm At the base of the ventricular septum the septal wall was defective for a distance of 1.2 cm and in this region the septal wall was depressed downward approximately 0 6 cm The superior portion of the ventricular septum deviated to the right to such an extent that the pulmonary orifice over rode the septal defect by a few millimeters From the upper margin of the ventricular septum close to the defect, a muscular ridge extended forward to the outer wall of the right ventricle This ridge separated the aorta from the pulmonary artery Con equently the aorta arose entirely from the right ventricle and only the pulmonary orifice overlay the ventricular septum Thus the

* A warning to prevent repetition of this serious mistake is given on p 235.

tions, however, do not relieve the pulmonary hypertension Hence the prognosis remains guarded

Illustrative Case

CASE XI-1 P A W (Harriet Lane Home, No A-60186) White female Referred for diagnosis of her cardiac abnormality in 1947, at five and a half years of age.

History Cyanosis was noted at birth and had persisted throughout life A murmur was first heard at three weeks of age Growth and development were slow She sat alone at nine months and walked at two years At three years of age she frequently squatted to rest but soon outgrew the habit

Physical examination Temperature 37°C , pulse 120 per minute, respirations 30 per minute, height 110 cm , weight 15 6 kg , and blood pressure 100/80 mm of mercury

The child was an intelligent, moderately cyanotic, poorly developed girl who suffered from dyspnea at rest There was suffusion of the conjunctivae The lips and buccal mucous membranes were deeply cyanotic The fingers and the toes were cyanotic and clubbed The heart was slightly enlarged The rhythm was regular A systolic murmur of moderate intensity was audible over the precordium, no thrill could be felt The lungs were clear to percussion and auscultation The liver and spleen were not palpable The pulse in the femoral artery was of good quality

Laboratory data Red blood cell count 9 3 million, hemoglobin 23 5 gm , hematocrit 77 per cent, oxygen saturation of the arterial blood 57 per cent

Teleoroentgenogram The heart was slightly enlarged There was fullness of the pulmonary conus and the hilar markings were increased (see Figure XI-4) Upon fluoroscopy faint expansile pulsations were visible in the hilar shadows Delineation of the esophagus with a barium-opaque mixture showed a left aortic arch and no evidence of left auricular enlargement

Electrocardiogram This showed a normal sinus mechanism, sinus tachycardia, high P waves in Lead II, right axis deviation, and evidence of right ventricular hypertrophy (see Figure XI-5)

Clinical impression The clinical findings were characteristic of an Eisenmenger complex in that there was cyanosis, clubbing, and polycythemia the heart was slightly enlarged with X ray evidence of fullness of the pulmonary conus and increased hilar shadows, which upon fluoroscopy showed faint expansile pulsations Nevertheless, the fact that cyanosis dated from birth made us suspect some totally different malformation For this reason special studies were undertaken

Cardiac catheterization revealed an oxygen content of 14 7 volumes per cent in the right auricle, 20 6 volumes per cent in the right ventricle, and 25 0 volumes per cent in the pulmonary artery, whereas the oxygen content in the aorta was only 17 4 volumes

2 Taussig H B and R J Bing Complete transposition of the aorta and a levoposition of the pulmonary artery Am Heart J 37 551–559 1949

3 Abbott M E *Atlas of Congenital Cardiac Disease* New York American Heart Association 1936

4 Beuren A The differential diagnosis of the Taussig Bing heart from complete transposition of the great vessels with a posteriorly overriding pulmonary artery Circulation 21 1071–1087 1960

5 Schultz F B An unusual congenital cardiac anomaly with associated anomalous venous return sudden death American Practitioner and Digest of Treatment 3 616–622 1952 Philadelphia Lippincott

6 Baffes T G A new method for surgical correction of transposition of the aorta and pulmonary artery Surg Gynec & Obst 102 227–233 1956

pulmonary artery not only received blood from the right ventricle but also received blood directly from the left ventricle The pulmonary artery and its branches were greatly dilated The pulmonary orifice measured 5 8 cm in circumference and the main pulmonary artery above the ring measured 6 5 cm The left and the right main branches were approximately 4 0 cm in circumference The pulmonary wall was thicker than normal, its intima, however, was smooth The ductus arteriosus was closed Examination of the myocardium showed that the fibers were hypertrophied but that there were no infarcts and no thrombi The coronary arteries appeared to be normal The bronchial arteries were not enlarged

The lungs were air-containing and showed no evidence of pneumonia or pulmonary infarcts, all the pulmonary vessels were patent Microscopic examination of the lungs revealed occasional thrombi, some of which were in the process of recanalization Many of the small pulmonary arterioles showed diffuse, intimal proliferation which rendered these vessels extremely narrow The lesion appeared to be sufficient to account for the increased resistance in the pulmonary vascular bed In addition, the pulmonary alveoli showed areas of emphysema and areas of atelectasis

The liver showed marked congestion it weighed 1,916 gm The spleen was enlarged and congested, it weighed 140 gm There were many small accessory spleens. The kidneys were normal except for congestion each weighed 80 gm The cortex and medulla were well defined The pelvis and ureters were normal There was diffuse hemorrhage in the thymus

Final anatomical diagnosis Transposition of the aorta Dilatation and slight displacement of the pulmonary artery Ventricular septal defect Foramen ovale covered by a valve, but not completely sealed Dilatation and hypertrophy of the right ventricle Extreme thickening and intimal proliferation of the pulmonary arterioles and small pulmonary arteries Occasional recanalization of thrombi in the pulmonary arterioles Splenomegaly Patchy emphysema and atelectasis Acute congestion of the lungs and viscera Diffuse hemorrhage in the thymus

Comment This was the first malformation of its type which the author had studied Diagnosis was correct in that the aorta arose from the right ventricle and the pulmonary artery occupied its normal position The fact that the pulmonary artery could be levoposed was not known to the author but the possibility should have been considered, because the oxygen content of the blood in the pulmonary artery was higher than that in the right ventricle and was also significantly higher than that in the aorta

References

1 Pernkopf E Die Partielle Situs inversus der Eingeweide beim Menschen Ztschr f Anat u Entwcklngsgesch 79 577-752 1926

cle pumped out 30 cc of blood, part of the blood would go to the lungs and part to the lower extremities Let us suppose that 20 cc go to the lungs and 10 cc. to the lower extremities The 20 cc which are directed to the lungs will be returned to the left auricle and this volume of blood will be pumped out from the left ventricle into the aorta All the blood in the aorta goes to the head and the upper extremities This blood will be returned to the right auricle by the superior vena cava In addition, the 10 cc of blood which were pumped from the right ventricle through the ductus arteriosus to the lower extremities will be returned to the right auricle by the inferior vena cava Therefore the right auricle will receive a total of 30 cc This amount will pass into the right ventricle Then the cycle starts again The course of the circulation is shown in Diagram XII–1

PHYSIOLOGY OF THE MALFORMATION

In this malformation the right ventricle not only pumps the blood to the lungs but also to the trunk and the lower extremities Consequently the pressure in the pulmonary artery is the same as that in the lower extremities It follows that the pressure in the lungs is unduly high Therefore, although the lungs receive venous blood from the right ventricle there is marked pulmonary hypertension Furthermore inasmuch as the right ventricle pumps all the blood which goes to the lungs and also all the blood which goes to the trunk and the lower extremities the right side of the heart carries a greater load than the left

CLINICAL FINDINGS

The difference in cyanosis between the upper and the lower extremities is the feature of diagnostic importance The head and the upper extremities receive oxygenated blood and therefore are not cyanotic, whereas the lower extremities receive venous blood which is ordinarily destined to go to the lungs for oxygenation and therefore are cyanotic The line of demarcation of the cyanosis lies at the brim of the pelvis, as it does in cases of complete transposition of the great vessels and not high up on the shoulders, as in cases of pulmonary hypertension with persistent patency of the ductus arteriosus In the malformation under discussion the left subclavian artery usually arises from the ascending aorta and carries fully oxygenated blood Inasmuch as there is a complete interruption of the isthmus of the aorta, no blood from the pulmonary artery can enter the subclavian artery, hence the cyanosis appears at a low level If, however, the left subclavian artery arises from the descending aorta, the line of demarcation of the cyanosis will be high up across the shoulders (see Chapter XVIII, Section B)

mation, the blood from the left ventricle is directed by way of the aorta to the head and the upper extremities and the blood from the right ventricle is pumped into the pulmonary artery and through the ductus arteriosus into the descending aorta The condition is readily compatible with fetal life and places no strain on the fetal circulation (see Figure XII–2)

After birth, however, the situation is quite different The blood from the left ventricle is pumped out by way of the aorta to the head and the upper extremities and is returned by the superior vena cava to the right auricle and to the right ventricle, thence it is pumped into the pulmonary artery Part of the blood from the pulmonary artery passes into the lungs and part through the ductus arteriosus to the descending aorta and the lower extremities The blood from the lungs is returned to the left auricle and the left ventricle, the blood from the lower extremities is returned by way of the inferior vena cava to the right auricle

Consequently, with each cardiac cycle, the right side of the heart receives a greater volume of blood than does the left side For example, if the right ventri

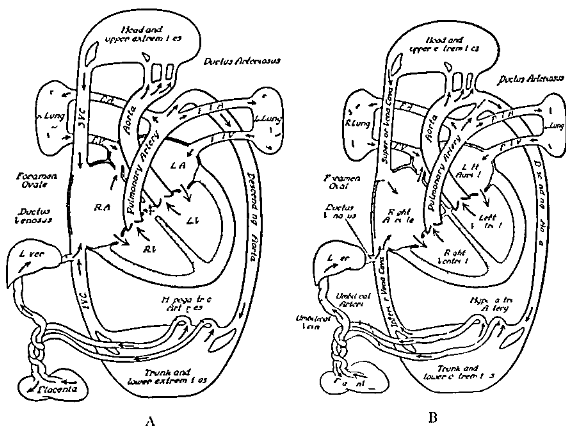


FIGURE XII–2 Fetal circulation (A) Complete interruption of the isthmus of the aorta and a persistent ostium atrioventriculare commune and (B) normal heart

cle pumped out 30 cc of blood, part of the blood would go to the lungs and part to the lower extremities Let us suppose that 20 cc. *go to the lungs and* 10 cc *to* the lower extremities The 20 cc which are directed to the lungs *will* be returned to the left auricle and this volume of blood will be pumped out from the left ventricle into the aorta All the blood in the aorta goes to the head and the upper extremities This blood will be returned to the right auricle by the superior vena cava In addition, the 10 cc of blood which were pumped *from the right ventri*cle through the ductus arteriosus to the lower extremities will be *returned to the right auricle* by the inferior vena cava Therefore the right auricle will receive a total of 30 cc. This amount will pass into the right ventricle Then the cycle starts again The course of the circulation is shown in Diagram XII–1

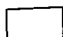
PHYSIOLOGY OF THE MALFORMATION

In this malformation the right ventricle *not only pumps* the blood to the lungs but also to the trunk and the *lower extremities* Consequently the pressure in the pulmonary artery is the same as that in the lower extremities It follows that the pressure in the lungs is unduly high Therefore, although the lungs receive venous blood from the right ventricle, there is marked pulmonary *hypertension* Furthermore inasmuch as the right ventricle pumps all the blood which goes to the lungs and also all the blood which *goes to the trunk* and the lower extremities, the right side of the heart *carries a greater* load than the left

CLINICAL FINDINGS

The difference in cyanosis between the upper and the lower extremities is the feature of diagnostic importance The head and the upper extremities receive oxygenated blood and therefore are not cyanotic, whereas *the lower* extremities receive venous blood which is ordinarily destined *to go to* the lungs for oxygenation and therefore are cyanotic The line of demarcation of the cyanosis lies at the brim of the pelvis, as *it does* in cases of complete transposition of the great vessels, and not high up *on the shoulders*, as in cases of pulmonary hypertension with persistent *patency of* the ductus arteriosus In the malformation *under* discussion the *left* subclavian artery usually arises from the *ascending* aorta and carries *fully* oxygenated blood Inasmuch as there is a *complete* interruption of *the* isthmus of the aorta no blood from the *pulmonary artery* can enter the subclavian artery hence the cyanosis appears at a low level If however the left subclavian artery arises from the descending aorta, the line of demarcation of the cyanosis will be high up across the shoulders (see Chapter XVIII, *Section* B)

Diagram XII–1


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

DIAGRAM XII–1

Absence of the aortic arch (complete interruption of the isthmus of the aorta) and a persistent ostium atrioventriculare commune The descending aorta is continuous with the pulmonary artery through the ductus arteriosus

The essential feature of this malformation is the complete interruption of the isthmus of the aorta The descending aorta is continuous with the pulmonary artery through the ductus arteriosus It follows that the pressure in the pulmonary artery is the same as that in the descending aorta Consequently there is marked pulmonary hypertension

It may be that a persistent ostium atrioventriculare commune or a ventricular septal defect is also an integral part of this malformation The significant changes in circulation however, depend primarily upon the complete interruption of the isthmus of the aorta

The blood from the right auricle flows into the right ventricle and is pumped out through the pulmonary artery to the lungs and through the ductus arteriosus into the descending aorta The blood from the lungs is returned to the left auricle and thence to the left ventricle and is pumped out through the aorta to the head and the upper extremities The blood from the head and the upper extremities is returned by the superior vena cava to the right auricle and the blood from the trunk and the lower extremities is returned by the inferior vena cava to the right auricle Therefore, with each cardiac cycle the right auricle receives all the blood which was pumped out from the left ventricle to the head and the upper extremities and also that part of the blood from the right ventricle which was pumped from the pulmonary artery through the ductus arteriosus to the trunk and the lower extremities The left auricle receives only that part of the blood from the right ventricle which was directed through the pulmonary artery to the lungs Therefore the pressure in the right auricle is greater than the pressure in the left auricle Some of the blood flows from the right auricle into the left auricle and some of the blood flows from the right ventricle into the aorta If the pressure on the left side of the heart rises the direction of the shunt will be reversed The circulation of the blood within the heart is similar to that in a persistent ostium atrioventriculare commune except that the pressure tends to remain higher in the right side of the heart as more blood is always returned to the right auricle than to the left auricle

Clinical diagnosis The most important diagnostic clue is the difference in cyanosis between the upper and the lower extremities The upper extremities receive oxygenated blood from the left side of the heart whereas the lower extremities receive venous blood from the right side of the heart because the descending aorta is continuous with the pulmonary artery Therefore the lower extremities will be cyanotic whereas the upper extremities will show no cyanosis The occurrence of a septal defect may or may not be an integral part of the malformation A murmur and a thrill if present are strongly suggestive of some additional defect

If the malformation is combined with a persistent ostium atrioventriculare commune, the difference in the intensity of the cyanosis between the upper and the lower extremities may be slight and will call for careful observation It is always best appreciated by placing the patient's hand beside his foot If the face and hands are of normal hue, the cyanosis of the feet may be overlooked Furthermore, if noted, it is likely to be attributed to cold or to a sluggish circulation The fact that the feet remain cyanotic when warm offers the clue to the correct diagnosis

The pulse is of good quality and of equal strength in the arm and the leg In contrast to coarctation of the aorta, this anomaly causes no obstruction to the flow of blood to the lower extremities The pulse in the lower extremities is normal

CARDIAC FINDINGS

The heart is slightly to moderately enlarged The enlargement affects the right side, it is due to the increased volume of blood which is returned to the right auricle and the right ventricle If the enlargement of the right ventricle is sufficiently great so that the right ventricle presses against the anterior chest wall, there will be left sided chest deformity

Murmurs may or may not be present

X RAY AND FLUOROSCOPIC FINDINGS

The heart is enlarged to the right and to the left Owing to the normal position of the pulmonary artery, the shadow at the base of the heart to the left of the sternum will be of normal contour Indeed, the pulmonary conus may be abnormally full

In the left anterior oblique position the enlargement of the right ventricle will be demonstrated by the projection of the anterior border of the cardiac silhouette toward the anterior chest wall The left ventricle will be seen to be small, it will require but little rotation of the patient for the left ventricle to clear the spinal column

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar precordial leads show evidence of right ventricular hypertrophy

DIAGNOSIS

The diagnosis is based upon the distribution of the cyanosis The head and

the upper extremities are of normal color, whereas the lower extremities are cyanotic because the descending aorta receives its blood from the pulmonary artery through the ductus arteriosus

DIFFERENTIAL DIAGNOSIS

The common error is the failure to appreciate the significance of the cyanosis of the feet If the head and the upper extremities are of good color, ordinarily little consideration is given to the circulation to the lower extremities Consequently the possibility of this abnormality is missed Even if cyanosis is noted, its significance is often not appreciated

TREATMENT

This abnormality could be corrected by the insertion of a graft between the ascending aorta and the descending aorta combined with the division of the ductus arteriosus

PROGNOSIS

The prognosis is usually poor If the condition should occur as an isolated anomaly successful surgery would restore the circulation to normal Usually, however the prognosis is determined by the associated anomaly, which may or may not be amenable to surgery

SUMMARY

The characteristic feature of a complete interruption of the isthmus of the aorta, in which the descending aorta becomes continuous with the pulmonary artery through the ductus arteriosus is that the cyanosis is confined to the lower extremities The pulse in both the upper and the lower extremities is strong The increased volume of blood which is pumped through the right auricle and the right ventricle produces enlargement of the right side of the heart, this in turn, may lead to left sided chest deformity

This malformation, if not associated with other anomalies, could be corrected by surgery

References

1 Abbott M E Personal communication

2 Congdon E D Transformation of the aortic arch system during the development of the human embryo Contrib Embryol 14 47-110 1922 Washington Carnegie Institution

CHAPTER XIII

DEFECTIVE DEVELOPMENT OF THE LEFT SIDE OF THE HEART

ATRESIA or marked hypoplasia of the aortic orifice is always associated with hypoplasia of the ascending aorta and also with defective development of the left ventricle In addition there may or may not be atresia of the mitral valve These conditions are discussed in Section A Section B presents an entirely different defect of the left ventricle, namely, a diverticulum of the left ventricle

A *Aortic Atresia or Marked Hypoplasia of the Aortic Orifice Combined with Hypoplasia of the Ascending Aorta*

This malformation is in a sense the counterpart of tricuspid atresia and a non functioning right ventricle Atresia or marked hypoplasia of the tricuspid valve is always associated with defective development of the right ventricle, the atresia of the pulmonary orifice is secondary to the failure of the right ventricle to function The aortic atresia and the marked hypoplasia of the ascending aorta are, however, the primary features of the malformation under discussion and the underdevelopment of the left ventricle and the mitral atresia are usually secondary to the abnormality of the aorta Thus in the former the primary abnormality concerns the tricuspid valve, which directs the blood to the right ventricle, whereas in the latter malformation the primary difficulty is the egress of blood from the left ventricle

NATURE OF THE MALFORMATION

The essential feature of this malformation is the atresia of the aortic valves or the marked hypoplasia of the aortic orifice combined with hypoplasia of the aorta up to the point of entrance of the ductus arteriosus Whether the aortic orifice is atretic or markedly hypoplastic, the underdevelopment of the aorta is always associated with an imperfect development of the left ventricle, either the left ventricle is absent or it is a thin walled, poorly developed chamber

The absence or marked hypoplasia of the left ventricle in turn affects the egress of blood from the left auricle When there is no left ventricle, the mitral

valve is always atretic and there is also a gross defect in the auricular septum When the aorta is markedly hypoplastic and the left ventricle is a relatively thin walled chamber, it is also common to find a small defect in the auricular septum In either case the main flow of blood to the systemic circulation is from the left auricle to the right auricle and thence to the right ventricle, from there the blood is pumped into the pulmonary artery and through the ductus arteriosus to the aorta The aorta, beyond the entrance of the ductus arteriosus, is usually of normal caliber The aorta, proximal to the entrance of the ductus arteriosus, always persists as a diminutive tube, and it is from the base of the aorta that the coronary arteries originate In cases of aortic atresia, the coronary arteries, and indeed the head and the upper extremities, receive their nutrition from the back flow of blood fom the ductus arteriosus to the base of the aorta When the aorta persists as a diminutive tube, the coronary arteries receive their supply of blood from the diminutive aorta in the normal manner Nevertheless, this malformation always causes real difficulty in the expulsion of blood from the left ventricle the main supply of blood to the systemic circulation is from the pulmonary artery through the ductus arteriosus Figure XIII–1 illustrates a case of aortic atresia mitral atresia and absence of the left ventricle Figure XIII–2 illustrates a case of marked hypoplasia of the aorta with a relatively thin walled left ventricle the aorta beyond the entrance of the ductus arteriosus is of normal caliber

Atresia of the aortic orifice and marked hypoplasia of the ascending aorta are always extremely serious Fortunately, although anatomically familiar, these conditions are clinically rare

The rarity of this malformation is in all probability due to the functional importance of the left ventricle during fetal life An embryo in which the left ventricle and aorta fail to develop can remain viable only if the function of the left ventricle is taken over by the right It is probable that in many instances the fetus dies at an early age In most instances the malformation is compatible with extra uterine life only for a short time

The condition is obviously more likely to be compatible with life when a defective left ventricle is combined with hypoplasia of the ascending aorta than when the aortic orifice is atretic Indeed, when the left ventricle is defective, the condition merges into that of pulmonary hypertension with persistent patency of the ductus arteriosus secondary to a severe left sided cardiac lesion In the older age group the signs and symptoms are primarily due to the pulmonary hypertension and the reversal of the flow of blood through the ductus arteriosus (see Chapter XVIII, Section B)

CHAPTER XIII

DEFECTIVE DEVELOPMENT OF THE LEFT SIDE OF THE HEART

ATRESIA or marked hypoplasia of the aortic orifice is always associated with hypoplasia of the ascending aorta and also with defective development of the left ventricle In addition there may or may not be atresia of the mitral valve These conditions are discussed in Section A Section B presents an entirely different defect of the left ventricle, namely, a diverticulum of the left ventricle

A *Aortic Atresia or Marked Hypoplasia of the Aortic Orifice Combined with Hypoplasia of the Ascending Aorta*

This malformation is in a sense the counterpart of tricuspid atresia and a non functioning right ventricle Atresia or marked hypoplasia of the tricuspid valve is always associated with defective development of the right ventricle, the atresia of the pulmonary orifice is secondary to the failure of the right ventricle to function The aortic atresia and the marked hypoplasia of the ascending aorta are, however, the primary features of the malformation under discussion and the underdevelopment of the left ventricle and the mitral atresia are usually secondary to the abnormality of the aorta Thus in the former the primary abnormality concerns the tricuspid valve, which directs the blood to the right ventricle, whereas in the latter malformation the primary difficulty is the egress of blood from the left ventricle

NATURE OF THE MALFORMATION

The essential feature of this malformation is the atresia of the aortic valves or the marked hypoplasia of the aortic orifice combined with hypoplasia of the aorta up to the point of entrance of the ductus arteriosus Whether the aortic orifice is atretic or markedly hypoplastic, the underdevelopment of the aorta is always associated with an imperfect development of the left ventricle, either the left ventricle is absent or it is a thin walled, poorly developed chamber

The absence or marked hypoplasia of the left ventricle in turn affects the egress of blood from the left auricle When there is no left ventricle, the mitral

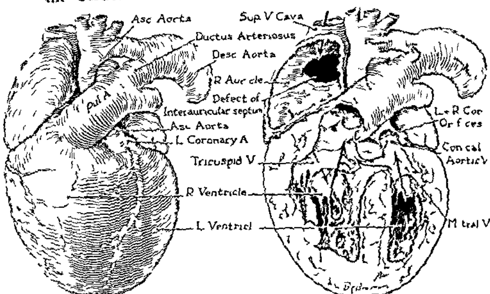


FIGURE XIII-2 Extreme aortic stenosis and hypoplasia of the ascending aorta with underdevelopment of the left ventricle, a high ventricular septal defect, and a gross defect in the auricular septum

COURSE OF THE CIRCULATION

During fetal life a considerable portion of the blood which enters the right auricle normally flows through the foramen ovale to the left auricle and thence to the left ventricle This blood is pumped through the aorta to the body of the fetus In cases of aortic atresia and a non functioning left ventricle, if the fetus is to remain alive, the work of the left ventricle must be taken over by the right ventricle Under such circumstances all the blood from the right auricle flows into the right ventricle and is pumped out into the pulmonary artery The major part of the blood which enters the pulmonary artery passes in the normal manner by way of the ductus arteriosus to the body of the fetus The volume of blood which is pumped through the ductus arteriosus is, however far greater than normal Virtually the entire work of the heart is performed by the right ventricle For this reason during intra uterine life the right ventricle undergoes great hypertrophy Indeed this is one of the few malformations in which the infant is born with enormous right sided cardiac enlargement Figure XIII-3 shows the fetal circulation in aortic atresia and Figure XIII-4 shows the fetal circulation in defective development of the left ventricle and marked hypoplasia of the ascending aorta

At birth with the expansion of the lungs there is an abrupt fall in the pul

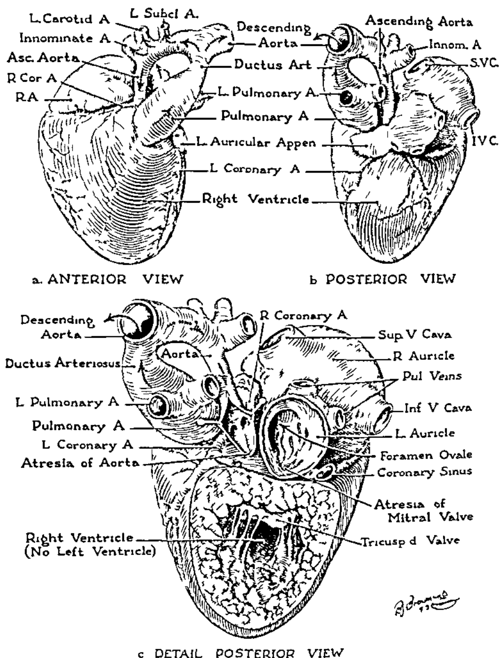


FIGURE XIII–1 Aortic atresia and marked hypoplasia of the ascending aorta, mitral atresia and absence of the left ventricle

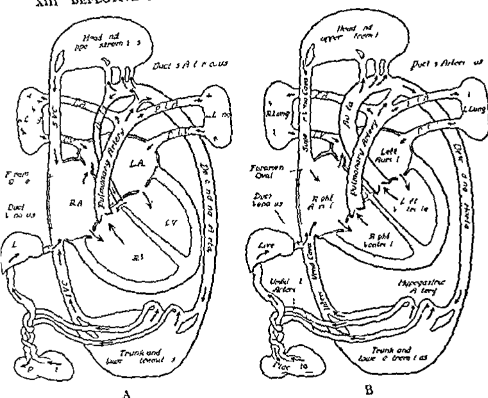


FIGURE XIII-4 Fetal circulation (A) Extreme aortic stenosis marked hypoplasia of the ascending aorta an underdeveloped left ventricle, and auricular and ventricular septal defects and (B) normal heart

non functioning, the blood cannot leave the left auricle in the normal manner All the blood from the left auricle must escape through a defect in the auricular septum to the right auricle

Thus the oxygenated blood which is returned from the lungs to the left auricle encounters difficulty in reaching the systemic circulation In the majority of other malformations cyanosis is due to the shunting of venous blood into the systemic circulation and thereby the oxygenated blood is diluted with venous blood In the malformation under discussion the body is primarily supplied with venous blood Furthermore, it is difficult for the oxygenated blood to reach the systemic circulation The course of the circulation is shown in Diagram XIII-1 Diagram XIII-2 shows the course of the circulation in a case of marked hypoplasia of the ascending aorta and defective development of the left ventricle

PHYSIOLOGY OF THE MALFORMATION

The right ventricle maintains the systemic circulation by pumping the blood

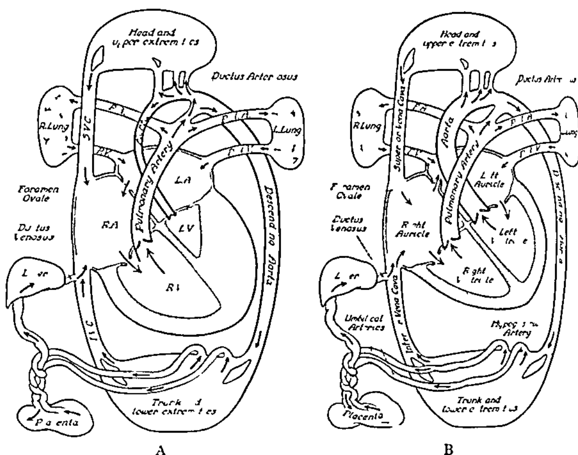


FIGURE VIII-3 Fetal circulation (A) Aortic atresia, mitral atresia, a non functioning left ventricle, and an auricular septal defect and (B) normal heart

monary pressure and consequently the blood in the pulmonary artery is directed to the lungs The sudden diversion of the blood to the lungs causes a cessation of the flow of blood through the ductus arteriosus to the aorta The consequence is that the systemic circulation, which has received its blood supply through this channel, is momentarily deprived of blood Not until the pressure in the pulmonary circulation becomes greater than that in the systemic circulation will the blood again flow through the ductus arteriosus to the aorta Then, and then only, will the systemic circulation be reestablished Moreover, upon the restoration of the circulation, the blood flows from the pulmonary artery to the aorta, the body is supplied with venous blood which is ordinarily destined to go to the lungs for oxygenation The blood which goes to the systemic circulation is returned by way of the superior and inferior venae cavae to the right auricle and thence it again passes to the systemic circulation The blood from the lungs is returned by way of the pulmonary veins to the left auricle If the left ventricle is absent or

Diagram XIII-1

Aortic atresia and marked hypoplasia of the ascending aorta combined with a patent ductus arteriosus mitral atresia a non functioning left ventricle and an auricular septal defect

The essential feature in this malformation is the atresia of the aorta at its base. The left ventricle does not function The left ventricle is either entirely absent or represented by a diminutive chamber filled with a blood clot. The mitral valve is completely atretic or markedly hypoplastic In addition there is always a defect in the auricular septum and the ductus arteriosus is patent. The blood supply to the systemic circulation is from the pulmonary artery through the ductus arteriosus The coronary arteries arise from the base of the aorta in the normal fashion but receive no blood from the left ventricle The blood supply to the myocardium is from the back flow of blood from the transverse arch of the aorta

The blood from the right auricle passes into the right ventricle and is pumped out by way of the pulmonary artery to the lungs and through the ductus arteriosus to the systemic circulation The blood from the lungs is returned to the left auricle. Inasmuch as there is an atresia of the mitral valve and the left ventricle does not function, the only way for the blood to escape from the left auricle is through the defect in the auricular septum to the right auricle Thence it passes to the right ventricle and is pumped out into the pulmonary artery Inasmuch as no blood is pumped from the left ventricle into the aorta, the pressure in the systemic circulation is low and blood flows from the pulmonary artery through the ductus arteriosus into the aorta This is the only way for the blood to reach the systemic circulation The coronary arteries arise from the base of the aorta and are fed by the blood which flows back from the ductus arteriosus to the ascending aorta and down to the base of the aorta The greater the volume of blood which reaches the lungs for oxygenation the greater is the deprivation of the systemic circulation At best the systemic circulation is supplied with a mixture of venous and arterial blood from the right ventricle. The circulation is very inefficient. The blood from the systemic circulation is returned in the normal manner by the superior and inferior venae cavae to the right auricle and to the right ventricle. The right ventricle pumps the blood not only to the lungs but also to the systemic circulation Hence there is always severe pulmonary hypertension Great hypertrophy of the right ventricle results

The blood in both the right auricle and the right ventricle is a mixture of the venous blood returned from the systemic circulation and of the oxygenated blood returned from the lungs With the complete expansion of the lungs the blood in the pulmonary artery is directed to the lungs and although a greater volume of blood is returned to the left auricle, the blood is directed away from the systemic circulation until the pressure in the pulmonary circulation exceeds that in the systemic circulation

Diagram XIII-1



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

from the pulmonary artery through the ductus *arteriosus* to the aorta Consequently the pressure in the pulmonary artery is the same as that in the systemic circulation Hence there is always pulmonary hypertension The only blood which the body ever receives is the blood which is pumped from the pulmonary artery through the ductus arteriosus to the aorta It is, however, important to remember that the blood in the pulmonary artery contains the oxygenated blood which has been returned by the pulmonary veins to the left auricle and shunted through the auricular defect to the right auricle, where it mixes with the venous blood returned by the superior and inferior venae cavae This mixture of arterial and venous blood flows into the right ventricle and is pumped out into the pulmonary artery and through the ductus arteriosus to the systemic circulation In the absence of an auricular defect, great difficulty is encountered in the expulsion of blood from the left auricle The increased pressure in the left auricle increases the resistance in the lungs and increases still further the work required of the right ventricle

CLINICAL FINDINGS

Cyanosis is always intense The baby is blue at birth In contrast to almost all other malformations the first breaths of life do nothing to relieve the cyanosis Indeed the establishment of respiration directs the blood to the lungs and away from the systemic circulation The only blood which the systemic circulation ever receives is the blood from the pulmonary artery which is ordinarily destined to go to the lungs for oxygenation Moreover the oxygenated blood which is returned from the lungs to the left auricle encounters difficulty in reaching the body Inasmuch as the aortic orifice is atretic and the entire systemic circulation is maintained through the ductus arteriosus, cyanosis is of uniform distribution

Dyspnea is severe

The pulse is so weak that it is frequently impalpable When palpable, it is of equal strength in the arm and the leg

The blood pressure is low

Engorgement of the liver and *edema* occur early

CARDIAC FINDINGS

The heart is enormously enlarged to the right and to the left Indeed, hypoplasia of the ascending aorta combined with defective development of the left ventricle is virtually the only malformation which causes the heart to be greatly enlarged at birth Figure XIII-5 shows the size of the heart in an infant twenty

Cyanosis is intense The condition is rarely compatible with life for more than a few days

Clinical diagnosis is based upon the early occurrence of intense cyanosis, weak pulses, great cardiac enlargement with forceful heart sounds, and right sided cardiac failure X ray or fluoroscopic examination shows that the enlargement is primarily due to the enlargement of the right ventricle In the anterior posterior position there is great enlargement of the pulmonary conus and in the left anterior-oblique position the right ventricle may be seen to extend to the anterior chest wall The condition of the infant remains critical throughout its brief life Death usually occurs within the first weeks of life

DIAGRAM XIII-2

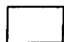
Extreme aortic stenosis and marked hypoplasia of the ascending aorta combined with an underdeveloped left ventricle a patent ductus arteriosus and auricular and ventricular septal defects

This malformation closely resembles that of aortic atresia The aorta, however, remains as a diminutive tube which may arise from the thin walled left ventricle or may over ride both ventri les In any event the main circulation to the body is by way of the right side of the heart the systemic circulation receives its blood from the pulmonary artery through the ductus arteriosus Consequently the systolic pressure in the right ventricle must b equal to the systemic pressure Hence there is a marked pulmonary hypertension

The blood from the right auricle flows into the right ventricle By far the greater part of the blood is pumped out into the pulmonary artery, thence the blood flows to the lungs and through the ductus arteriosus to the systemic circulation The blood from the lungs is returned in the normal fashion by the pulmonary veins to the left auricle A small portion of the blood flows from the left auricle into the left ventricle and is pumped out through the stenosed aortic orifice into the diminutive aorta The greater part of the blood from the left auricle flows through the defect in the auricular septum to the right auricle, where it mixes with the blood which is returned by the superior and inferior venae cavae to the right auricle Virtually all the blood from the right auricle flows into the right ventricle and is pumped out into the pulmonary ar tery When the diminutive aorta over rides the ventricular septum, a small volume of blood from the right ventricle can be pumped directly into the aorta, nevertheless the aorta is so small that the systemic circulation receives but a minimal amount of blood from the diminutive aorta The coronary arteries, however, arise from the base of the aorta in the normal fashion and receive their blood from the small volume of blood which is pumped into the diminutive ascending aorta

Clinical diagnosis The clinical findings are essentially the same as those of aortic atresia The infant is born with great right sided cardiac enlargement the x ray shows marked fullness of the pulmonary conus, cyanosis is intense. Cardiac failure occurs early the condition is compatible with life for only a few days

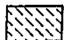
DIAGRAM XIII–2



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

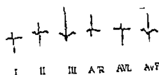
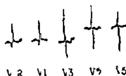


FIGURE XIII-6 Defective development of the left ventricle and hypoplasia of the ascending aorta (same patient as in Figure XIII-5) Infant



ular septum is small, there may be enlargement of the left auricle The duration of life is usually too short to permit great enlargement of the left auricle The baby is often too feeble to swallow barium and consequently no evidence can be obtained concerning the size of the left auricle

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar leads show evidence of marked right ventricular hypertrophy The T waves may be of low amplitude (see Figure XIII-6)

SPECIAL TESTS

The condition of the infant is generally too critical to permit more than a brief examination

Cardiac catheterization would reveal a left to-right shunt at the auricular level and systemic pressure in both the right ventricle and the pulmonary artery It would probably be possible to catheterize the descending aorta through the ductus arteriosus

Angiocardiography would reveal simultaneous opacification of the descending aorta and the pulmonary artery

DIAGNOSIS

The diagnosis is based upon the findings of intense cyanosis, enormous right sided cardiac enlargement which dates from birth, the forcefulness of the heart sounds and the weakness of the pulses, and early signs of right sided cardiac failure

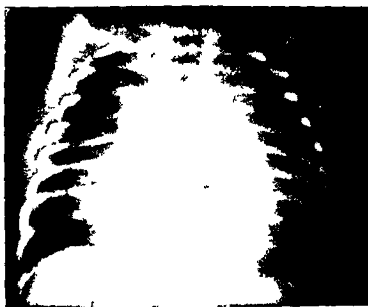


FIGURE XIII–5 Defective development of the left ventricle and hypoplasia of the ascending aorta (same patient as in Figure XIII–6) Infant twenty four hours of age

four hours old In this instance the enormous size of the cardiac shadow, detected when the infant was twelve hours of age, had raised the question of a mediastinal tumor *The heart sounds* are forceful *Murmurs* may or may not be present *Cardiac failure* occurs early and within the first two or three days of life the liver becomes engorged and extends to the umbilicus

X RAY AND FLUOROSCOPIC FINDINGS

Fluoroscopic examination shows great enlargement of the right auricle, of the right ventricle, and of the pulmonary conus In addition, the superior vena cava is often dilated, consequently the shadow at the base of the heart is abnormally wide Upon rotation of the infant s head to the left, the pulmonary artery can be visualized in its normal position The lungs remain relatively clear because the pulmonary vascular bed opens up slowly The heart may be so enormous that only in the left anterior oblique position is it possible to ascertain that the increased size of the heart is primarily due to great enlargement of the right ventricle In this position the anterior border of the cardiac silhouette extends to the anterior chest wall Examination in the right anterior-oblique position reveals little The cardiac shadow appears to fill the entire chest

The size of the left auricle depends upon the size of the auricular defect If there is mitral atresia in addition to the aortic atresia, and the defect in the auric

and a foramen ovale guarded by a valve may cause early signs of distress At birth, however, the heart is normal in size, the enlargement is barely detectable at two days of age

Pulmonary hypertension with reversal of the flow of blood through the ductus arteriosus, when caused by a severe left sided cardiac lesion, may be due to defective development of the left ventricle and marked hypoplasia of the ascending aorta If the condition is not extreme and the left ventricle supplies the blood to the head and the upper extremities, the clinical syndrome is that of pulmonary hypertension and a reversed ductus, in which the trunk and the lower extremities are more cyanotic than the head and the upper extremities (see Chapter XVIII, Section B) When there is atresia of the aortic orifice, the entire circulation is maintained by the right ventricle The work of the right ventricle is greatly increased and the heart is greatly enlarged, cyanosis is intense and of uniform distribution

Truncus arteriosus may be confused with aortic atresia, especially when the circulation to the lungs is by way of the bronchial arteries Under such circumstances cyanosis appears early and is associated with great cardiac enlargement The contour of the heart, however, is different In a truncus arteriosus, the shadow at the base of the heart to the left of the sternum is concave, whereas in aortic atresia the pulmonary conus is abnormally prominent Furthermore, in the right anterior-oblique position the sharp angulation of the right ventricle from the aorta to the anterior chest wall is seen only in a truncus arteriosus (see Chapter XIV) the exaggeration of the pulmonary conus in an aortic atresia causes a diffuse bulge along the anterior margin of the cardiac silhouette

TREATMENT

There is no effective treatment An aortic atresia and a marked hypoplasia of the ascending aorta, combined with a poorly developed left ventricle, constitute such a severe abnormality that surgical correction is impossible

PROGNOSIS

The prognosis is hopeless The condition of the infant remains precarious throughout its brief life Cardiac failure occurs early The malformation is usually compatible with life for only a few days

SUMMARY

The outstanding features of aortic atresia are intense cyanosis, great cardiac enlargement and the early onset of cardiac failure The heart sounds are force

The fluoroscopic findings give the clue to the diagnosis The tremendous size of the right ventricle indicates that the right, not the left ventricle, is the chamber of functional importance in the maintenance of the systemic circulation The enlargement of the pulmonary conus shows that the pulmonary artery is given off the right ventricle in the normal fashion These two findings together indicate that the blood from the right ventricle is pumped through the pulmonary artery to the systemic circulation It follows that the ductus arteriosus must be widely patent and of functional importance in the direction of the blood from the pulmonary artery to the aorta In order for the blood to flow from the pulmonary artery through the ductus arteriosus to the aorta, the pressure in the systemic circulation must be lower than that in the pulmonary circulation Therefore the flow of blood from the left ventricle into the aorta must be meager From these facts it is deduced that the primary malformation is an aortic atresia or marked hypoplasia of the aortic orifice combined with hypoplasia of the ascending aorta

Inasmuch as this malformation is always associated with underdevelopment of the left ventricle, the blood cannot leave the left auricle in the normal manner Consequently there must be some defect in the auricular septum The clinical manifestations of the auricular septal defect vary according to its size and may be difficult to determine If the defect is large, the thin walled, readily distensible right auricle bears the brunt of the increased auricular pressure This, too, can be detected upon fluoroscopy by the distention of the superior vena cava If the defect is small, the left auricle becomes enlarged and causes backward displacement of the esophagus

DIFFERENTIAL DIAGNOSIS

Aortic atresia calls for differentiation from other anomalies which place great strain on the right side of the heart and from other conditions which are fatal in early infancy

Total anomaly of the pulmonary venous return in the terminal stage may be confused with aortic atresia, because in both instances in early infancy the patient may be intensely cyanotic The history is usually of aid, as a baby with a total anomaly of the venous return is seldom intensely cyanotic from birth except when all the pulmonary veins drain into the hepatic vein, under such circumstances the heart is phenomenally small

"Pure" pulmonary stenosis may cause cyanosis and cardiac failure in the neonatal period Furthermore, at this age the heart is but slightly enlarged The infant with "pure" pulmonary stenosis usually responds promptly to digitalis

Complete transposition of the great vessels with a closed ventricular septum

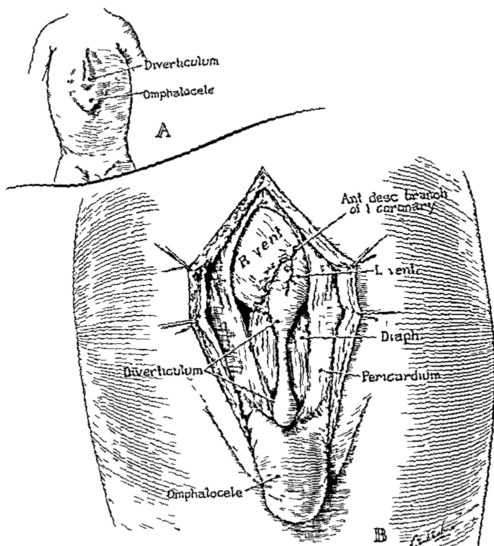


FIGURE XIII-7 Diverticulum of the left ventricle. Case XIII-1

heart with increased pulmonary blood flow The pulsating vessel in the abdominal wall was thought to be a diverticulum of the left ventricle but the possibility of an aberrant vessel arising from the descending aorta was also considered

Special tests Angiocardiography was performed in the usual manner The right side of the heart filled normally Then the left side of the heart and the aorta were visualized There was slight opacification below the diaphragm at the time the dye entered the left ventricle but it was not clearly delineated The descending aorta was clearly delineated and it was obvious that no abnormal vessel arose therefrom

ful Murmurs may or may not be present The pulse is weak but, if palpable, is of equal intensity in the arm and the leg Fluoroscopic examination shows that the enlargement is primarily right sided and that the pulmonary conus is abnormally full The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

The condition calls for differentiation from other malformations which cause cardiac failure in the neonatal period and are compatible with life for only a short time A total anomaly of the venous return is sometimes confused with this malformation Pulmonary stenosis with an intact ventricular septum and complete transposition of the great vessels with a closed ventricular septum may cause cardiac failure in the neonatal period Congenital mitral atresia may closely simulate this malformation There is as yet no treatment The prognosis is hopeless Death usually occurs within the first few days

B *Diverticulum of the Left Ventricle*

A diverticulum which extends from the apex of the left ventricle through the diaphragm into the abdomen is a rare abnormality, but several cases have been reported [1 3] It is usually associated with an omphalocele and is frequently associated with tricuspid atresia The diagnosis is made by the pulsating mass in the upper abdomen The following case report illustrates the condition

Illustrative Case

CASE VIII-1 Baby K (Harriet Lane Home, No A-96602) White male First seen in the Cardiac Clinic in September, 1952, at two months of age because of a heart murmur

Physical examination The patient was well nourished and well-developed and had good color The head and neck showed no abnormalities Examination of the abdomen revealed a large omphalocele (see Figure VIII-7) and, within its upper portion, a vessel which pulsated vigorously and extended up to the diaphragm The vessel felt like a thick walled artery about 3 mm in diameter It was possible to slide one s fingers beneath the distal end of this vessel It was also possible to constrict the vessel in its mid portion, when this was done both the proximal and the distal segments pulsated vigorously

Examination of the heart revealed slight cardiac enlargement The heart sounds were of good quality The pulmonic second sound was accentuated and there was a harsh precordial systolic murmur

Fluoroscopy revealed slight cardiac enlargement and increased hilar shadows

Clinical impression The baby obviously had a congenital malformation of the

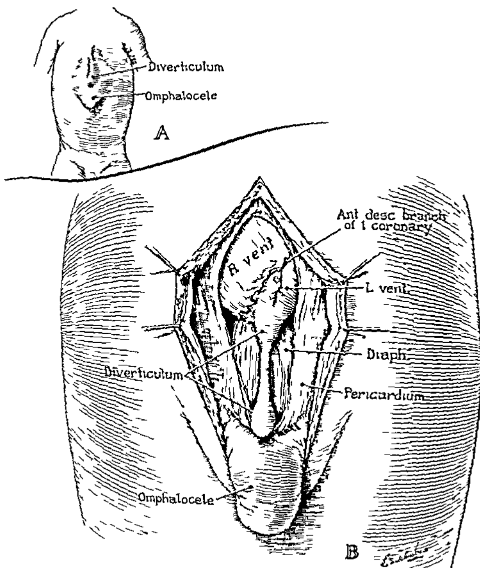


FIGURE XIII-7 Diverticulum of the left ventricle Case XIII-1

heart with increased pulmonary blood flow The pulsating vessel in the abdominal wall was thought to be a diverticulum of the left ventricle but the possibility of an aberrant vessel arising from the descending aorta was also considered

Special tests Angiocardiography was performed in the usual manner The right side of the heart filled normally Then the left side of the heart and the aorta were visualized There was slight opacification below the diaphragm at the time the dye entered the left ventricle but it was not clearly delineated The descending aorta was clearly delineated and it was obvious that no abnormal vessel arose therefrom

Clinical diagnosis Congenital malformation of the heart and a diverticulum of the left ventricle

Disposition Although in 1952 surgical correction of the cardiac malformation was not possible, it was hoped that removal of the diverticulum would strengthen the left ventricle Therefore surgery was recommended

Treatment Surgical correction was attempted but, as soon as the diverticulum was dissected and a clamp was placed on it, the heart stopped and could not be restarted

Autopsy No 23910 (limited, performed by Dr Morgan Berthrong) The external examination of the infant revealed that the umbilicus was absent and that there was a protrusion of the anterior abdominal wall which formed a hernial pouch 1 5 cm below the tip of the diaphragm This mass measured 6 x 9 cm There was a surgical incision which extended down the mid portion of the mass When the surgical incision was opened, it was apparent that a portion of the abdominal mass was continuous with the pericardial sac A prolongation of the pericardial sac extended beneath the xiphoid into the subcutaneous tissues of the anterior abdominal wall The anterior portion of the diaphragm was not attached to the xiphoid or to the costal cartilages at the mid line The musculature of the upper anterior abdominal wall was missing and the finger like projection of the diverticulum extended from the left ventricle through the defect In addition there was a small nodule of myocardial muscle found free in the subcutaneous fat of the upper abdominal wall

The heart was enormously enlarged the enlargement was due solely to the greatly dilated right auricle and the greatly hypertrophied right ventricle The superior and inferior venae cavae entered normally into the right auricle The foramen ovale was anatomically patent The tricuspid valve was normal and opened into a greatly hypertrophied right ventricle The wall of the right ventricle was 1 cm in thickness in many places There was a large defect in the ventricular septum The pulmonary valve and pulmonary ring were normal The pulmonary artery was greatly dilated The pulmonary veins entered a small left auricle, the mitral valve and the left ventricle were normal in size Extending downward from the apex of the left ventricle there was a diverticulum consisting of a long, narrow chamber The orifice of the chamber was only 3 mm in diameter The chamber measured 6 5 cm in length and varied in width as shown in Figure xiii-8 The myocardium of the chamber was thin, it measured approximately 1 to 2 mm in thickness except at the tip There was a high ventricular septal defect The aorta, however, arose normally from the left ventricle and did not over ride the defect The ascending aorta measured 1.2 cm in diameter The left common carotid artery arose anomalously from the innominate artery The aorta between the left subclavian artery and the point of entrance of the ductus arteriosus was only 7 mm in diameter Beyond the ductus arteriosus the aorta again increased to 9 mm in diameter



FIGURE XIII–8 Diverticulum of the left ventricle
Case XIII–1

The lungs were grossly normal Microscopic sections of the lungs showed extreme muscular hypertrophy of the smallest pulmonary arterioles with tiny lumina No intimal lesions were seen Thus the small pulmonary arterioles appeared similar to systemic arterioles The diverticulum showed normal endocardium and myocardium The liver was normal

Final anatomical diagnosis Congenital malformation of the heart ventricular septal defect, patent foramen ovale, patent ductus arteriosus and diverticulum of the left ventricle coarctation of the aorta proximal to the ductus arteriosus origin of the left common carotid artery from the innominate artery pulmonary arteriosclerosis right ventricular hypertrophy congenital defect in the pericardium congenital malformation of the diaphragm diastasis recti abdominis with ventral hernia containing cardiac diverticulum myocardial fibers in abdominal subcutaneous tissue ventral surgical incision with attempted removal of cardiac diverticulum

Comment The diagnosis of diverticulum of the left ventricle was correct The severity of the pulmonary hypertension undoubtedly contributed to the cardiac difficulty and increased the risk of operation, though it was not the apparent cause of death

References

1 Swyer A J, I H Mauss and P Rosenblatt Congenital diverticulosis of left ventricle Am J Dis Child 79 111–114, 1950

2 Skapinker, S Diverticulum of the left ventricle of the heart A M A Arch Surg 63 629–634, 1951

3 Potts, W J Congenital diverticulum of the left ventricle, case report Surgery 33 301–307, 1953

CHAPTER XIV

TRUNCUS ARTERIOSUS AND HEMI-TRUNCUS ARTERIOSUS

A TRUNCUS arteriosus means that there is a single great vessel which directs the blood to the systemic circulation and to the lungs Hemi truncus arteriosus is the name given to the condition in which one pulmonary artery arises directly from the aorta, as it does in a true truncus arteriosus, and the other pulmonary artery arises normally from the right ventricle The former condition is discussed in Section A, the latter in Section B

A *Truncus Arteriosus*

A truncus arteriosus is a single great vessel of abnormally large caliber which combines the features of both great vessels This vessel receives all the blood from both ventricles and directs the blood to the systemic circulation and to the lungs The circulation to the lungs is by way of the pulmonary artery which arises directly from the base of the single great vessel, or there are no pulmonary arteries and the circulation to the lungs is established through the bronchial arteries or some anomalous vessels of the collateral circulation which may be given off from almost any portion of the aorta The coronary arteries arise from the base of the common trunk, which continues as the aorta and supplies the systemic circulation in the normal manner

Formerly, this malformation was classified as a true truncus arteriosus only provided that the orifice was guarded by a valve with four semilunar cusps and pulmonary arteries were given directly off the single great vessel The condition was termed a pseudo truncus arteriosus" or a truncus aorticus if a vestigial pulmonary artery was found, or if the valve guarding its orifice had less than four cusps, even though there was but a single great vessel and the circulation to the lungs was by way of the bronchial arteries The occurrence of four semilunar cusps has been generally discarded as an essential criterion Humphreys,[1] in a careful analysis of an extensive series of cases expressed the opinion that a pseudo truncus arteriosus represents an earlier developmental arrest in the formation of the great vessels than does a true truncus arteriosus In a true' truncus arteriosus the pulmonary artery has developed but has failed to become separated from the aorta in a pseudo truncus arteriosus the pulmonary artery has not yet reached the stage at which it joins the truncus

A review of the development of the great vessels from the primitive aortic arches is necessary to understand the relation of the two conditions and the *fundamental nature of the resulting malformation*

EMBRYOLOGY

The aorta is derived from the fourth branchial arch and the pulmonary artery from the sixth Normally the sixth left branchial arch distal to the pulmonary artery persists as the ductus arteriosus

Both the fourth branchial arch, which forms the aorta, and the sixth arch, which forms the pulmonary artery, originate from the aortic sac, whence the *aortic trunk leads to the bulbus cordis*

A so-called "true" truncus arteriosus occurs if the aortic septum fails to develop within the primitive aortic sac, under such conditions the pulmonary artery arises directly from the common trunk Often the only remnant of the aortic septum present is a fold in the wall of the aorta just above the point where the main pulmonary artery branches off from the common trunk In other cases the formation of the aortic septum has progressed to the stage at which the left and right pulmonary arteries arise from the single great vessel through a common orifice This common orifice or common vessel may be given off from the posterior wall of the ascending aorta or higher up on the arch of the aorta (see the diagram of the truncus arteriosus in Figure XIV–1) In the rare instances in which the pulmonary arteries arise directly from the truncus arteriosus, the ductus arteriosus is normally formed, in most instances, however, it is absent *Inasmuch as the function of the ductus arteriosus is to direct the* blood from the pulmonary artery to the aorta, it serves no purpose when the pulmonary artery and the aorta arise from a common trunk Therefore, since the ductus arteriosus is of no functional importance, it is not surprising to find that it may atrophy and disappear quite as readily as does the distal end of the sixth right branchial arch

The condition is also considered a "true" truncus arteriosus when there is a single large truncus guarded by a valve with four semilunar cusps and no vestige of a pulmonary artery is found Under such circumstances the circulation to the lungs is through the bronchial arteries or other pathways of the collateral circulation Such vessels are usually minute and the pulmonary blood flow is meager

A pseudo truncus arteriosus represents an arrest in the *formation of the sixth* branchial arch which is of such a nature that it fails to meet the aortic sac Usually, when the proximal end of the sixth branchial arch fails to meet the aortic

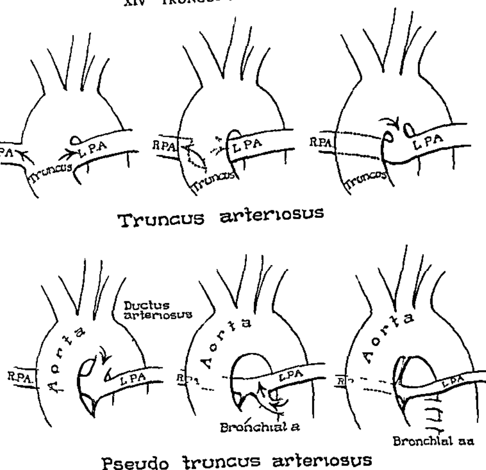


FIGURE XIV–1 Truncus arteriosus with pulmonary vessels arising from the truncus and pseudo truncus arteriosus

sac, the distal end fails to meet the dorsal aorta Consequently, not only does the pulmonary artery fail to meet the aorta in the normal fashion, but the ductus arteriosus is absent Under such circumstances a vestigial pulmonary artery exists, it leads to the lungs but does not connect with the heart or the aorta and consequently it carries no blood Under such circumstances, as in a true truncus arteriosus in which the pulmonary artery is absent, the only way for the blood to reach the lungs is by way of the collateral circulation, which is usually established through the bronchial arteries Thus the circulation to the lungs may be identical in the two conditions

Humphreys showed that in a very real sense a true truncus arteriosus and a pseudo truncus arteriosus could exist together She cited a case in which one pulmonary artery arose directly from the aorta and the other pulmonary artery

Comment The diagnosis of diverticulum of th severity of the pulmonary hypertension undoubtedl culty and increased the risk of operation, though death

References

1 Swyer, A J, I H Mauss and P Rosenblatt Cc Am J Dis Child 79 111–114, 1950
2 Skapinker, S Diverticulum of the left ventri 63 629–634 1951
3 Potts, W J Congenital diverticulum of the lei 307, 1953

cs
'eft
com-
m the
rta (see
stances in
riosus, the
it is absent
od from the
monary artery
uctus arteriosus
it may atrophy
h right branchial

sus when there is a
r cusps and no vestige
the circulation to the
of the collateral circu
blood flow is meager
he formation of the sixth
meet the aortic sac Usu
rch fails to meet the aortic

the coronary arteries has not been seen by the author, either at operation or at autopsy, or at least it has not been recognized as such

Although in most instances, it is relatively easy to locate the anomalous vessels as they arise from the aorta or its branches, it is extremely difficult to trace their entrance into the lungs Occasionally, as mentioned above, a large vessel can be seen to enter the pulmonary artery In most instances, however, it is well nigh impossible to demonstrate the exact manner in which the vessels of collateral circulation anastomose with the pulmonary vascular bed

Collister et al [4] have made careful studies of the development of the pulmonary vascular bed They believe that when there is pulmonary atresia the pathways of the collateral circulation are laid down during intra uterine life This must certainly be true in the case of a truncus arteriosus in which the entire circulation to the lungs is by way of anomalous vessels If it is true in one instance, it is certainly reasonable to believe that it is true in another The concept implies that the ultimate effectiveness of the collateral circulation is dependent upon the vascular channels laid down during intra uterine life

It has been the author s experience that some patients with a pseudo truncus arteriosus have been greatly helped by a systemic pulmonary anastomosis and that other patients have developed thromboses far out in the lungs distal to the anastomosis Thus it appears that in some instances the pulmonary arteries are able to direct blood to the lungs and that in other instances the vestigial pulmonary artery does not open normally into the pulmonary vascular bed It is the author s impression that if the vessels of the collateral circulation open into the pulmonary arteries, then the distal pulmonary vascular bed is normal and can direct the blood to the lungs, if, however, the vessels of the collateral circulation enter the lungs far out in the capillary bed, or connect with the bronchial arterial system then the vestigial pulmonary artery lacks its normal connection with the pulmonary capillary bed and at best it can carry only a small volume of blood to the lungs

Furthermore even upon direct observation at operation, it may be impossible to determine whether or not there is a vestigial pulmonary artery Consequently the differentiation of a true truncus arteriosus from a pseudo truncus arteriosus, on the basis of the existence of a rudimentary pulmonary artery, can be established only at autopsy Because of this and because there are all gradations of both types of truncus arteriosus, these two conditions are considered as variants of the same malformation namely a truncus arteriosus

Additional evidence that these two conditions represent closely related de

was a blind tube, that is, it failed to connect with the heart or the aorta, the circulation to the lung with the blind pulmonary artery was established by way of the bronchial arteries The author has seen two cases in which one pulmonary artery arose from the aorta and the other arose in the normal fashion from the right ventricle (see Section B)

Christeller,[2] in his detailed study of a series of cases of pulmonary atresia, found that when the ductus arteriosus did not develop or did not remain patent there were eight other possible pathways by which the collateral circulation might be established

1 The anterior bronchial arteries, that is, the superior bronchial arteries which arise from the arch of the aorta
2 The posterior bronchial arteries, which arise from the posterior wall of the descending aorta or from the intercostal arteries
3 The anterior mediastinal arteries, which arise from the internal mammary arteries
4 The posterior mediastinal arteries which arise from the posterior wall of the aorta
5 Branches from the esophageal arteries, which arise from the posterior mediastinal arteries
6 Branches from the pericardial arteries which arise either from the esophageal arteries or from the posterior mediastinal arteries
7 Anomalous branches which may arise from the coronary arteries of the heart
8 Anomalous arteries which may arise from the aorta or occasionally from the subclavian arteries, from the diaphragmatic arteries, or from the pericardial arteries

Enlargement of the anterior and posterior mediastinal vessels has been commonly observed at operation In almost every instance the collateral circulation over the anterior chest wall is evident and the mediastinal vessels are large and numerous Furthermore, the posterior mediastinal vessels may be so numerous as to cause increased hilar shadows In many patients large anomalous vessels coursing across the lungs have been seen at operation In one instance a large vessel which apparently arose from the descending aorta opened directly into the inferior wall of the pulmonary artery, as shown in the middle diagram of pseudo truncus arteriosus in Figure XII-1 Dr Paul White[3] has seen one patient in whom at operation the circulation to the left lung apparently came from a large vessel which arose from the descending aorta The author has seen one similar instance in which at autopsy the main vessel of collateral circulation was found to arise from the aorta below the level of the diaphragm In addition, she has also seen one child who suffered from hematemesis which, in all probability, was due to hemorrhage from esophageal varices secondary to the enlargement of the esophageal arteries Collateral circulation from the pericardial arteries or

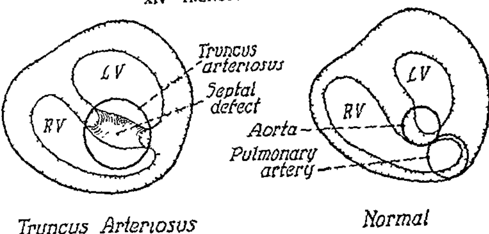


FIGURE XIV–2 Truncus arteriosus and normal heart

ductus arteriosus is usually absent when present, it is seldom of functional importance

A truncus arteriosus is often associated with an arrest in the development of the heart itself Defects in the auricular septum are common, occasionally the ventricular septum fails to develop and there is but a single ventricle Such conditions, however, represent additional anomalies, they are not part and parcel of the truncus arteriosus A single auricle or even a single ventricle does not greatly alter the circulation, because all the blood is pumped out into the common vessel Therefore, regardless of whether or not venous and arterial blood are mixed in the auricle or in the ventricle, complete admixture occurs in the truncus arteriosus Furthermore, there is no separation of the work of the two sides of the heart Either a common ventricle or two ventricles pump the blood through a single great vessel to both the systemic and the pulmonary circulation

COURSE OF THE CIRCULATION

During fetal life the circulation to the lungs is minimal, the main flow of blood is by way of the single vessel to the body of the fetus The blood is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle The blood which goes to the lungs is returned in the normal fashion to the left auricle Nevertheless, inasmuch as the truncus arteriosus arises more from the right ventricle than from the left, the work of the right ventricle is increased (see Figures XIV–7 and 8)

After birth the establishment of respiration opens the pulmonary vascular bed and thereby some blood from the truncus arteriosus is directed to the lungs

velopmental arrests is that they cause virtually the same alteration in the size and the shape of the heart The fundamental contour of the heart is determined during intra uterine life During fetal life the circulation to the lungs is relatively unimportant, consequently the work of the heart is virtually the same whether the circulation to the lungs is from the pulmonary artery or from the bronchial arteries Moreover, when the circulation to the lungs is by way of the bronchial arteries, existence of a rudimentary pulmonary artery is of no functional importance In both cases, the two ventricles pump the blood through the single great vessel to the body of the fetus

Moreover, in either a pseudo truncus arteriosus or a "true" truncus arteriosus, the pulmonary blood flow may be excessive or it may be markedly reduced Therefore, instead of an anatomical separation of the two conditions, the author prefers a physiological differentiation on the basis of the volume of the pulmonary blood flow, which may be increased, normal, or decreased

At the stage in the development of the heart when there is but a single great vessel, there are but a single auricle and a single ventricle, the single great vessel arises from the primitive bulbus cordis As the aortic septum develops, the common trunk shifts from its position relative to the bulbus cordis to assume a more posterior position above the left ventricle When the development of the great vessel is arrested, the truncus arteriosus arises from both ventricles, but usually more from the right than from the left

NATURE OF THE MALFORMATION

A truncus arteriosus is a single great vessel of unusually large caliber which combines the features of both great vessels The common truncus over rides the ventricular septum and receives the blood from both ventricles, hence a high ventricular septal defect is an integral part of the malformation (see Figure XIV–2) The orifice of the truncus is guarded by a valve with two, three, or four semilunar cusps The coronary arteries arise from the base of the great vessel The pulmonary artery arises either from the posterior wall of the truncus (see Figure XIV–3) or from its base as in the case reported by MacGilpin,[6] which is shown in Figures XIV–4 and 5, or else the pulmonary artery is absent and the circulation to the lungs is established by way of the bronchial vessels or by some anomalous vessels as illustrated in Figure XIV–6 In all instances the blood from both ventricles is pumped out into the common arterial trunk and thence it flows through the arterial pathways to the systemic circulation and to the lungs The heart is slightly to moderately enlarged Both ventricles are thick walled The



FIGURE XIV-4 Truncus arteriosus with a single pulmonary artery arising from the base of the truncus (same patient as in Figures XIV-3 18)

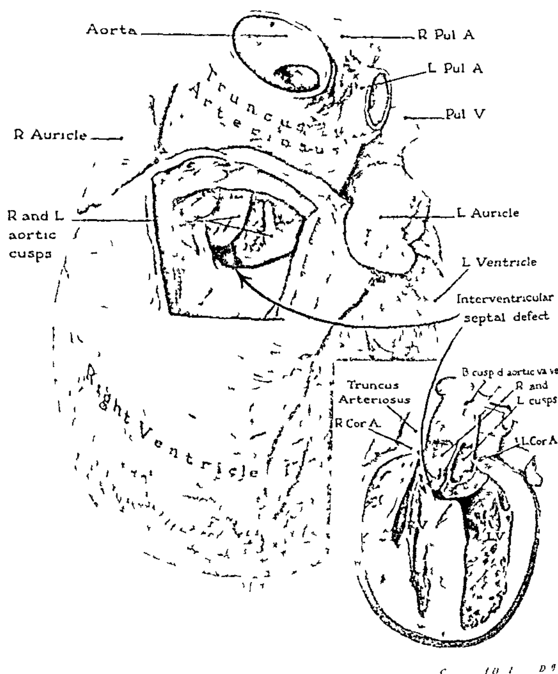


FIGURE XIV–3 Truncus arteriosus with the pulmonary arteries arising from the main truncus and a high ventricular septal defect

The single vessel over rides the septum and receives blood from both ventricles The left and the right coronary arteries arise from the base of the *aorta behind the semilunar cusps*



FIGURE XIV-4 Truncus arteriosus with a single pulmonary artery arising from the base of the truncus (same patient as in Figures XIV-5 18)

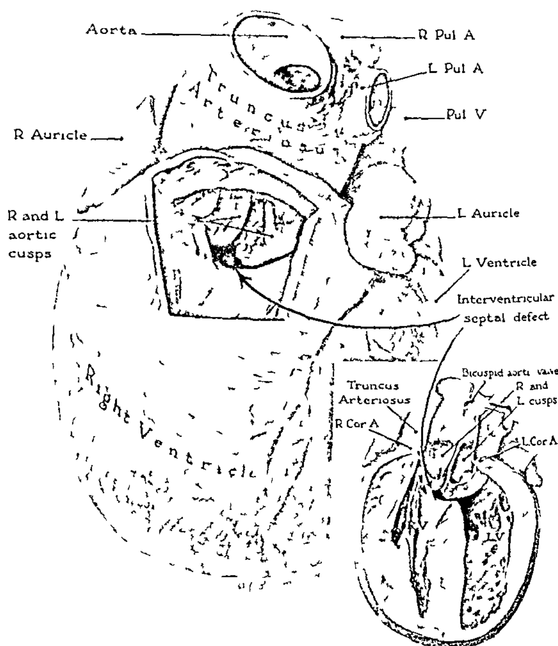


FIGURE XIV-3 Truncus arteriosus with the pulmonary arteries arising from the main truncus and a high ventricular septal defect

The single vessel over rides the septum and receives blood from both ventricles The left and the right coronary arteries arise from the base of the aorta behind the semilunar cusps

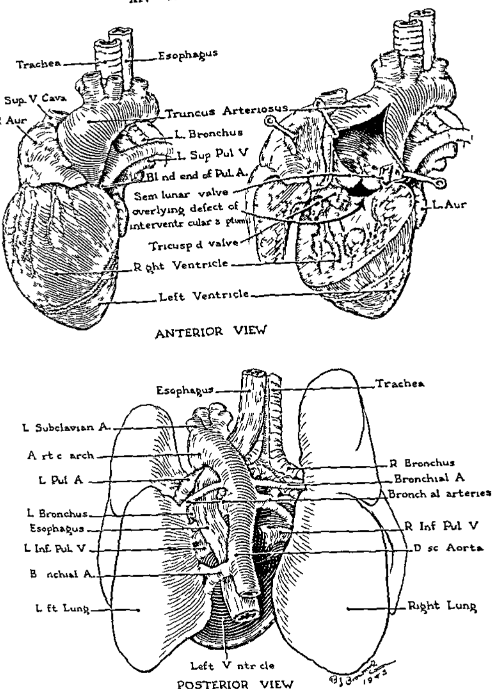


FIGURE XIV–6 Pseudo truncus arteriosus (same patient as in Figure XIV–20)

The anterior view shows the truncus arteriosus over riding the ventricular septum The posterior view shows the enlarged bronchial arteries and the esophagus caught between the bronchial arteries and displaced to the left

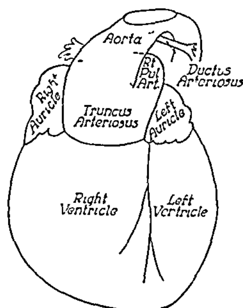


Figure xiv–5 Truncus arteriosus with a single pulmonary artery arising from the base of the truncus (same patient as in Figure xiv–4)

If the pulmonary arteries arise from the truncus, a large volume of blood is directed to the lungs, where it is oxygenated, and returned in the normal manner by the pulmonary veins to the left auricle, thence it flows to the left ventricle The remainder of the blood in the truncus arteriosus is directed to the body and is returned by the superior and inferior venae cavae to the right auricle, thence it flows to the right ventricle The blood from both ventricles is pumped out into the common trunk There the cycle starts again Under such circumstances a large volume of oxygenated blood is mixed with the venous blood which is returned from the body to the right side of the heart, therefore cyanosis is minimal or absent (see Diagram xiv–1)

When the pulmonary artery fails to meet the aorta and the ductus arteriosus is absent, the only possible way for blood to reach the lungs is through the bronchial arteries or some anomalous vessels Although these pathways of collateral circulation enlarge, they are seldom sufficient to permit adequate circulation to the lungs The volume of blood which flows to the lungs varies with the size and number of these pathways When these are few or of very small caliber, the pulmonary blood flow is extremely meager Only a small volume of oxygenated blood is returned to the left auricle and the left ventricle Hence only a small volume of oxygenated blood is mixed with a large volume of venous blood, consequently cyanosis is intense (see Diagram xiv–2)

Thus a truncus arteriosus may have excessive, normal, or reduced pulmonary blood flow Nevertheless, the course of the circulation in all instances is basically the same

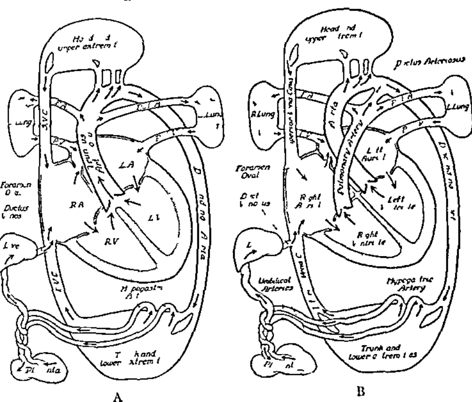


FIGURE XIV–8 Fetal circulation (A) Truncus arteriosus with the pulmonary arteries arising from the main truncus and (B) normal heart

nary artery is related to volume of the pulmonary blood flow When relatively small vessels are given off the descending aorta, the pulmonary pressure is usually abnormally low

CLINICAL FINDINGS

The presence or absence of cyanosis varies with the adequacy of the pulmonary blood flow Cyanosis is minimal or absent in infancy and childhood when the pulmonary arteries arise directly from the common trunk Under such circumstances a large volume of blood reaches the lungs for oxygenation and a large volume of oxygenated blood is returned to the left side of the heart Furthermore, the blood is directed into the pulmonary arteries under systemic pressure Consequently, if the pulmonary vascular bed opens up in the normal fashion, there is real danger of excessive flow to the lungs and the depletion of the systemic circulation This phenomenon is the probable explanation of the high mortality rate in the neonatal period when the pulmonary arteries arise

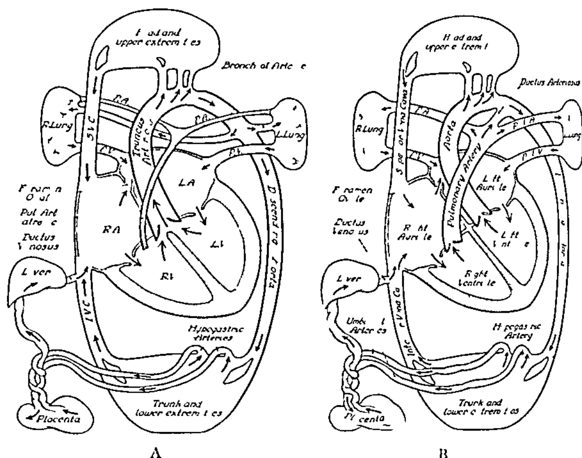


FIGURE XIV-7 Fetal circulation (A) Truncus arteriosus with circulation to the lungs by way of the bronchial arteries and (B) normal heart

PHYSIOLOGY OF THE MALFORMATION

In this malformation all the blood to the body and to the lungs is pumped out through the common truncus Consequently the same admixture of venous and arterial blood is directed to the two circulations The height of the pressure in the lesser circulation depends upon the size and the position of the vessels which direct the blood to the lungs The pressure in the ascending aorta is a combination of the force of the actual expansion of the aorta and that of the forward propulsion of the blood, whereas in the descending aorta the expansile force is no longer present and the pressure is caused solely by the force of the forward propulsion of the blood[7, 8] Consequently, if large pulmonary arteries are given off the ascending portion of the truncus arteriosus, the pressure in the pulmonary artery is the same as that in the systemic circulation, under such circumstances pulmonary hypertension is severe If, however, large vessels are given off the descending aorta, the hemodynamics are similar to those which occur with a huge ductus arteriosus, namely, the height of the pressure in the pulmo-

DIAGRAM XIV–1

Truncus arteriosus with the pulmonary arteries arising from the main truncus

The essential feature of this malformation is that there is but a single great vessel which combines the features of both great vessels, it receives the blood from both ventricles and directs blood to the systemic circulation and to the lungs The coronary arteries arise from the base of this single vessel and the pulmonary arteries originate as branches of the main trunk Both auricles are normally formed and both ventricles are normally formed except for a high ventricular septal defect This is inevitable as the truncus arteriosus arises from both ventricles, hence it over rides a high ventricular septal defect Both ventricles are abnormally thick

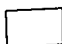
The blood from the right auricle flows into the right ventricle and that from the left auricle flows into the left ventricle. From both ventricles the blood is pumped out into the common arterial trunk and flows both to the body and through the pulmonary arteries to the lungs The blood from the head and the upper extremities is returned by the superior vena cava to the right auricle and that from the trunk and the lower extremities is returned by the inferior vena cava to the right auricle The blood which is pumped into the pulmonary arteries flows in the normal manner to the lungs where it is oxygenated and the oxygenated blood is returned by the pulmonary veins to the left auricle There the cycle starts again Inasmuch as the pulmonary arteries are of normal size a large volume of oxygenated blood is returned to the left auricle and to the left ventricle This blood is pumped out into the common arterial trunk where it mixes with venous blood from the right side of the heart Inasmuch as a large volume of oxygenated blood is mixed with the normal venous blood, cyanosis is minimal or absent.

Clinical diagnosis If the pulmonary arteries are large and are given off at the base of the truncus the pressure in the pulmonary arteries is the same as that in the aorta there is marked pulmonary hypertension and consequently there is no continuous murmur

Frequently both pulmonary arteries are given off from a single orifice, which creates a mild functional pulmonary stenosis under such circumstances either an early diastolic murmur is audible at the base of the heart or a continuous murmur is audible over the lungs The electrocardiogram usually shows a right axis deviation and evidence of either right ventricular hypertrophy or combined ventricular hypertrophy

Diagram XIV-1

Head and upper extremities
R Lung
L Lung
S.V.C.
P.A.
Pul A.
PV
Pul V
Truncus Arteriosus
Descending
Aorta
L.A.
L.V.
R.A.
R.V.
Coronary-Artery
I.V.C.
Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

Diagram XIV–1

Truncus arteriosus with the pulmonary arteries arising from the main truncus

The essential feature of this malformation is that there is but a single great vessel which combines the features of both great vessels, it receives the blood from both ventricles and directs blood to the systemic circulation and to the lungs The coronary arteries arise from the base of this single vessel and the pulmonary arteries originate as branches of the main trunk Both auricles are normally formed and both ventricles are normally formed except for a high ventricular septal defect This is inevitable as the truncus arteriosus arises from both ventricles, hence it over rides a high ventricular septal defect Both ventricles are abnormally thick

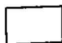
The blood from the right auricle flows into the right ventricle and that from the left auricle flows into the left ventricle. From both ventricles the blood is pumped out into the common arterial trunk and flows both *to the body and through the pulmonary* arteries to the lungs The blood from the head and the upper extremities is returned by the superior vena cava to the right auricle and that from the trunk and the lower extremities is returned by the inferior vena cava to the right auricle The blood which is pumped into the pulmonary arteries flows in the normal manner to the lungs where it is oxygenated and the oxygenated blood is returned by the pulmonary veins to the left auricle There the cycle starts again Inasmuch as the pulmonary arteries are of normal size a large volume of oxygenated blood is returned to the left auricle and to the left ventricle This blood is pumped out into the common arterial runk where it mixes with venous blood from the right side of the heart Inasmuch as a large volume of oxygenated blood is mixed with the normal venous blood, cyanosis is minimal or absent.

Clinical diagnosis If the pulmonary arteries are large and are given off at the base of the truncus the pressure in the pulmonary arteries *is the same as that in the aorta* there is marked pulmonary hypertension and consequently there is no continuous murmur

Frequently both pulmonary arteries are given off from a single orifice which creates a mild functional pulmonary stenosis under such circumstances either an early diastolic murmur is audible at the base of the heart or a continuous murmur is audible over the lungs The electrocardiogram usually shows a right axis deviation and evidence of either right ventricular hypertrophy or combined ventricular hypertrophy

DIAGRAM XIV–2

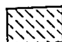
Head and upper extremities
Pulmonary Arteries
Bronchial Arteries
R Lung
L Lung
S.V.C.
PV
Pul V
Pseudo-Truncus Arteriosus
L.A
R.A
L.V
R.V
Descending Aorta
Remnant of Pul Art
I.V.C.
Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

Diagram XIV–2

Pseudo truncus arteriosus

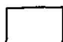
In this malformation there is but a single great vessel which receives the blood from both ventricles and directs it to the systemic circulation and to the lungs The pulmonary arterie, however do not arise from this great vessel The circulation to the lungs is by way of the bronchial arteries or anomalous vessels of the collateral circulation Both auricles and both ventricles are normally formed, inasmuch as the common arterial trunk arises from both ventricles a high ventricular septal defect is inevitable The coronary arteries arise at the base of the truncus arteriosus behind the semilunar cusps Inasmuch as the pulmonary artery does not connect with the truncus arteriosus or with the heart the only pathway by which the blood can reach the lungs is through the bronchial arteries There may be one or two abnormal superior bronchial arteries which arise from the arch of the aorta or some of the posterior bronchial arteries may be abnormally large

The blood from the right auricle flows into the right ventricle and is pumped out by way of the common arterial trunk to the body and through the bronchial arteries to the lungs The blood from the head and the upper extremities is returned by the superior vena cava to the right auricle and that from the trunk and the lower extremities is returned by the inferior vena cava to the same auricle The blood in the bronchial arteries flows to the lungs and the oxygenated blood is returned in the normal manner by the pulmonary veins to the left auricle Thence the blood flows to the left ventricle and is pumped out into the truncus arteriosus There the cycle starts again

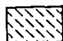
Clinical diagnosis The patient usually shows persistent cyanosis but is not severely limited The heart is enlarged there is a concave curve at the base of the heart to the left of the sternum a prominent high aortic knob and decreased hilar markings, and an absence of the hilar comma A continuous murmur is audible over one or both lungs the intensity of the murmur is inversely proportional to the intensity of the cyanosis The electrocardiogram usually shows a right axis deviation and commonly shows evidence of combined hypertrophy

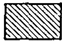
Diagram XIV-2

Head and upper extremities
Pulmonary Arteries
Bronchial Arteries
R Lung
L Lung
S.V.C.
PV
Pul V
Retrotracheal Anastomosis
LA
RA
LV
RV
Remnant of Pul Art
IVC
Descending Aorta
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

become obliterated; hence most infants with a truncus arteriosus do not suffer from attacks of paroxysmal dyspnea Moreover, the vessels of collateral circulation tend to increase in size as the patient grows and *consequently* the circulation to the lungs may gradually show slight improvement

Exercise tolerance of the individual varies *directly* with the volume of the pulmonary blood flow When the pulmonary arteries arise directly *from* the aorta, there is adequate or excessive pulmonary blood flow and the *individual* can do virtually as much as any normal person In contrast to this, when the circulation to the lungs is by way of the *minute* vessels of collateral circulation, the patient s activity is extremely limited An intermediate stage between these two extremes is the most common of all Namely, the collateral circulation is moderately well developed Under such circumstances, although the pulmonary blood flow is reduced, it is constant Furthermore, since the arteries which carry the blood to the lungs arise directly from the aorta, any increase in the systemic pressure increases the circulation to the lungs The result is that most patients can walk considerable distances and do not squat when tired In this malformation the systemic pressure regulates the pressure under which the blood flows to the lungs Indeed, the pulmonary *blood flow remains relatively constant unless* there is a collapse of the systemic circulation It is important to remember that in a patient whose pulmonary blood flow is extremely meager a sudden drop in the systemic pressure may be fatal

The pulse is forceful and of equal strength in the arm and the leg Arterial pulsations are frequently conspicuous in the vessels of the neck

The blood pressure is, however, usually normal Indeed, the diastolic pressure is usually higher than might be expected from the quality of the pulse

CARDIAC FINDINGS

The heart is slightly to moderately enlarged The enlargement mainly involves the right ventricle which may be so great as to cause left sided chest deformity

The second sound at the base is loud and pure It may be better heard to the left of the sternum than to the right it is, however, never reduplicated

A harsh systolic murmur is frequently audible over the precordium and may be widely transmitted throughout the chest When the murmur is intense, a *thrill* develops at an early age the thrill is of maximal intensity over the sternum at the base of the heart

A continuous murmur which is best heard over the lungs is the most charac

directly from the base of the truncus Once a balance is established, the patient usually does well for a number of years Unfortunately when the pulmonary arteries arise from the base of the truncus, these vessels receive blood under systemic pressure As in all such instances, initially the pulmonary bed opens up slowly and the hypertension is compensatory Over the years, however, intimal changes occur and become progressively more severe Eventually the vascular changes may be so great as to reduce the volume of blood which reaches the lungs for oxygenation and thus they increase the volume of venous blood directed to the systemic circulation to such an extent that the patient develops cyanosis and polycythemia Therefore, terminally, most adults with this malformation show varying degrees of cyanosis, clubbing, and polycythemia

Cyanosis almost always dates from birth when the pulmonary artery is absent and the circulation to the lungs is through the bronchial arteries or anomalous vessels of the collateral circulation Only rarely are these vessels sufficiently large so that there is adequate circulation to the lungs Usually these pathways are extremely small in comparison with the size of the normal pulmonary artery and the pulmonary blood flow is proportionately reduced Under such circumstances, only a small volume of blood goes to the lungs and a large volume of blood goes to the body, hence only a small volume of oxygenated blood is returned to the left auricle and a large amount of venous blood is returned to the right auricle. Regardless of whether or not there is a gross defect in the auricular septum, all the blood returned to the auricles is pumped out into the common great vessel, hence the greater the reduction of the pulmonary blood flow, the smaller is the volume of oxygenated blood directed to the systemic circulation and the lower is the oxygen saturation of the arterial blood Under such circumstances cyanosis is readily apparent at birth and polycythemia develops in early infancy

Dyspnea varies with the adequacy of the pulmonary blood flow Nevertheless, the volume of the pulmonary blood flow, regardless of whether it is great or small, is related to the systemic pressure, therefore it is relatively constant For this reason, although polypnea may be marked, respirations are not labored and attacks of paroxysmal dyspnea are rare Indeed, paroxysmal dyspnea occurs only in those rare instances in which the ductus arteriosus constitutes the principal pathway by which the blood can reach the lungs As the ductus periodically constricts during the process of closure, the circulation to the lungs is abruptly reduced, under such circumstances, an infant may suffer from attacks of paroxysmal dyspnea Usually, however, the circulation to the lungs is maintained by small arteries which arise directly from the aorta These vessels do not tend to

ter heard posteriorly than anteriorly The murmur may be maximal over the base of the heart to the left of the sternum, under such circumstances it requires differentiation from the murmur of persistent patency of the ductus arteriosus The murmur produced by a truncus arteriosus differs from that associated with patency of the ductus arteriosus in that it has a humming quality and varies in intensity with respiration, it is usually better heard with the breath held in full expiration Ordinarily the murmur is not very loud and is limited to one side of the chest It may be localized in almost any place, depending on the location of the anomalous vessels to the lungs Thus the murmur may be audible over a limited area, front, back, or in the axilla Such a murmur frequently escapes detection unless carefully sought for when listening over the lungs

A thrill is palpable only when the murmur is excessively loud When a thrill does occur, it is not localized over the pulmonary area but is widely transmitted throughout the lungs and may be readily palpable in both axillae A continuous thrill, however is the exception not the rule

X RAY AND FLUOROSCOPIC FINDINGS

In infancy the contour of the heart is distinctive, especially when the pulmonary arteries are absent In the anterior posterior position the heart is seen to be greatly enlarged Owing to the absence of the normal pulmonary artery, the shadow cast by the pulmonary conus is absent, the upper margin of the cardiac shadow to the left of the sternum is concave The ventricular shadow extends nearly to the axilla and the apex of the heart is upturned owing to the enlargement of the right ventricle (see Figures xiv–9 and 10)

In the right anterior-oblique position the upper margin of the shadow cast by the right ventricle makes an abrupt angle with the aorta and extends horizontally outward to the anterior chest wall It is so straight that it has the appearance of a shelf or wall (see Figures xiv–11 and 12) If Puck could sit on the edge of a non functioning right ventricle, Humpty Dumpty could balance on this wall

In the left anterior-oblique position the shelf may or may not be apparent, depending on the thickness of the right ventricle Inasmuch as the truncus arises more from the right ventricle than from the outflow tract, there is usually an angulation between the truncus arteriosus and the right ventricle In Figure xiv–11 the patient is not rotated sufficiently far to show the enlargement of the right ventricle but the anterior displacement of the esophagus by the retroesophageal vessels is clearly visible (see below)

teristic of all findings in a truncus arteriosus The murmur is seldom present at birth but frequently develops during the early months of life It is depend n on the continuous flow of blood from the aorta through the vessels of the col lateral circulation to the lungs Thus it depends on the height of the systemic pressure, on the size of the vessel leading to the lungs, and on the pulmonary resistance

When the pulmonary arteries arise directly from the base of the heart, the pressure in these vessels is the same as that in the systemic circulation If the lungs expand rapidly, the pulmonary blood flow becomes excessive, when this occurs no continuous murmur is heard and the infant dies at an early age Often the orifice of the pulmonary artery is sufficiently small to break the high pressure in the truncus Under such circumstances, as the pulmonary vascular bed gradually opens, the difference in pressure between the systemic and the pul monary circulation becomes sufficiently great to cause a continuous murmur

If the vessels of the collateral circulation are minute, so little blood flows through these vessels that there will be no continuous murmur Fortunately, the vessels of the collateral circulation tend to increase in size with the growth of the individual Therefore it is not uncommon for a continuous murmur to appear during infancy As previously mentioned, in rare instances when the main pathway of the collateral circulation is by way of the ductus arteriosus, as the ductus arteriosus undergoes obliteration the infant may suffer from attacks of paroxysmal dyspnea The author has had the opportunity to examine two such patients during episodes of paroxysmal dyspnea In both instances the continu ous murmur disappeared during the attack of paroxysmal dyspnea and upon the return of the murmur the infant s color immediately improved and dyspnea subsided

Usually, however, the vessels are of moderate size—small enough to break the force of the systemic pressure and yet sufficiently large to permit blood to flow continuously to the lungs The intensity of the continuous murmur is di rectly proportional to the volume of blood which reaches the lungs Conse quently, if the patient shows minimal or no cyanosis and correspondingly little incapacity, there is a loud continuous murmur If there is moderate cyanosis and some limitation of activity, the murmur may be localized over one lung or over one portion of one lung, whereas if there is intense cyanosis and marked incapacity, there may be no murmur

The murmur may be of maximal intensity in almost any location If the mur mur is loud, it is usually well heard over one or both lungs It is frequently bet

ter heard posteriorly than anteriorly The murmur may be maximal over the base of the heart to the left of the sternum, under such circumstances it requires differentiation from the murmur of persistent patency of the ductus arteriosus The murmur produced by a truncus arteriosus differs from that associated with patency of the ductus arteriosus in that it has a humming quality and varies in intensity with respiration, it is usually better heard with the breath held in full expiration Ordinarily the murmur is not very loud and is limited to one side of the chest It may be localized in almost any place, depending on the location of the anomalous vessels to the lungs Thus the murmur may be audible over a limited area front, back, or in the axilla Such a murmur frequently escapes de tection unless carefully sought for when listening over the lungs

A thrill is palpable only when the murmur is excessively loud When a thrill does occur it is not localized over the pulmonary area but is widely transmitted throughout the lungs and may be readily palpable in both axillae A continuous thrill, however is the exception not the rule

X RAY AND FLUOROSCOPIC FINDINGS

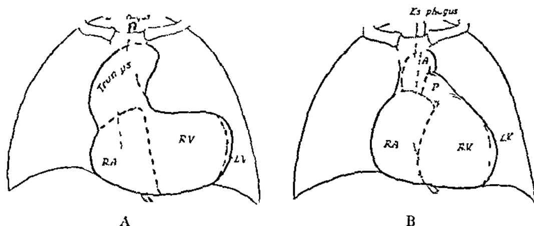
In infancy the contour of the heart is distinctive, especially when the pulmonary arteries are absent In the anterior posterior position the heart is seen to be greatly enlarged Owing to the absence of the normal pulmonary artery, the shadow cast by the pulmonary conus is absent, the upper margin of the cardiac shadow to the left of the sternum is concave The ventricular shadow extends nearly to the axilla and the apex of the heart is upturned owing to the enlarge ment of the right ventricle (see Figures XIV–9 and 10)

In the right anterior-oblique position the upper margin of the shadow cast by the right ventricle makes an abrupt angle with the aorta and extends hori zontally outward to the anterior chest wall It is so straight that it has the appear ance of a shelf or wall (see Figures XIV–11 and 12) If Puck could sit on the edge of a non functioning right ventricle, Humpty Dumpty could balance on this wall

In the left anterior-oblique position the shelf may or may not be apparent, depending on the thickness of the right ventricle Inasmuch as the truncus arises more from the right ventricle than from the outflow tract, there is usually an angulation between the truncus arteriosus and the right ventricle In Figure XIV–11 the patient is not rotated sufficiently far to show the enlargement of the right ventricle but the anterior displacement of the esophagus by the retro-esophageal vessels is clearly visible (see below)



FIGURE XIX-9 Truncus arteriosus (same patient as in Figures XIX-11, 21) Infant

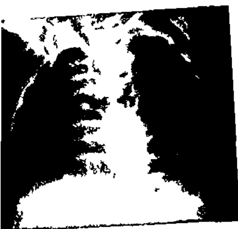


A B

FIGURE XIX-10 (A) Truncus arteriosus and (B) normal heart Infant

The truncus arteriosus is of abnormally large caliber and arches at an abnormally high level as shown in Figure XIX-9 and also in Figures XIX-13, 15, 19, and 24 In infancy the abnormally large aorta is not only conspicuous but also usually indents the esophagus The indentation of the esophagus can generally be demonstrated in the anterior posterior position and in both oblique positions upon its delineation with a radio-opaque mixture A right aortic arch is quite common with this malformation, as shown in Figures XIX-13 and 14

As the individual grows, the diaphragm descends and the heart comes to occupy a more vertical position in the chest than it did in infancy When this



Left anterior-oblique position



Right anterior-oblique position

FIGURE XIV–11 Truncus arteriosus (same patient as in Figure XIV–9) Infant Note the shelf like projection of the right ventricle

occurs the contour of the heart usually changes The right ventricle no longer projects forward so abruptly, consequently the contour of the heart comes to resemble that of a tetralogy of Fallot with a severe pulmonary stenosis (see Figures XIV–15 and 16) Under such circumstances the high aortic arch, in combination with an absence of the shadow cast by the normal pulmonary artery and its branches, gives the clue to the diagnosis

Absence of the shadow cast by the right and left branches of the main pulmonary artery may be striking Danelius[9] emphasized the absence of a hilar comma in cases of truncus arteriosus In the author s experience, not only is the

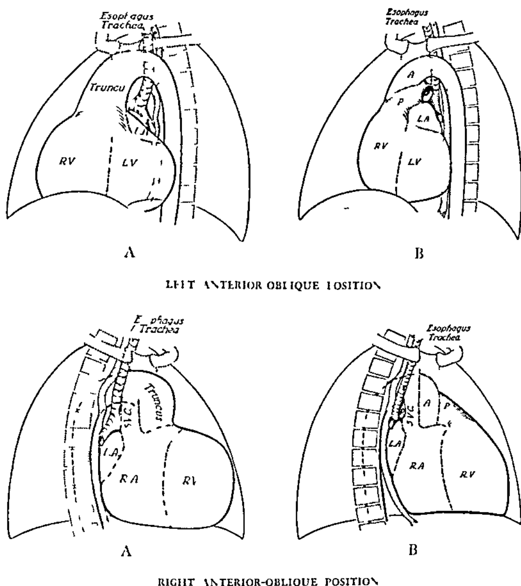


FIGURE XIV-12 (A) Truncus arteriosus and (B) normal heart Infant

hilar comma absent, but it is replaced by diffuse fine vascular markings which radiate from both hilar regions to the lungs

On the other hand, *if large pulmonary arteries are given directly off from the common trunk,* they receive a large volume of blood under systemic pressure Under such circumstances, although the shadow at the base of the heart has a concave curve, there may be a conspicuous hilar dance (see Figure XIV-17)

Occasionally one or both pulmonary arteries arise from the base of the truncus on the left, close to the origin of the normal pulmonary artery, as shown in Figures XIV-4 and 5 Under such circumstances, in contrast to the usual con

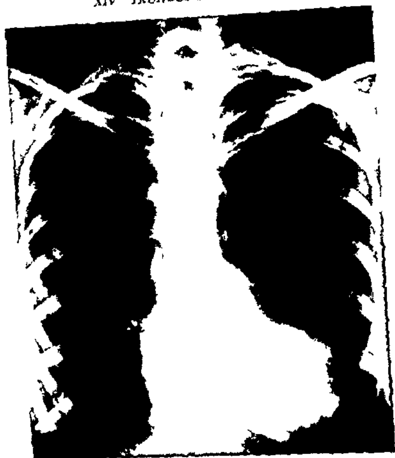


Figure XIV–13 Truncus arteriosus with a right aortic arch. Adult

cavity there may be fullness in the region of the pulmonary conus, as shown in Figure XIV–18

The occurrence of anomalous retro-esophageal vessels offers additional evidence that the circulation to the lungs is by way of collateral pathways The esophagus may be caught between anomalous bronchial arteries and displaced anteriorly or laterally as shown in Figures XIV–11 and 12 Indeed, the demonstration of retro-esophageal vessels in the lower thoracic region is of diagnostic significance, as it cannot be due to any abnormality of the vessels which arise from the aortic arch (see Chapter XXVI) In rare instances the esophagus may be caught by one or more of these anomalous vessels and displaced in an extremely bizarre manner as shown in Figure XIV–19

When the main circulation to the lungs is by way of the superior bronchial arteries these vessels may become greatly dilated In rare instances the increased

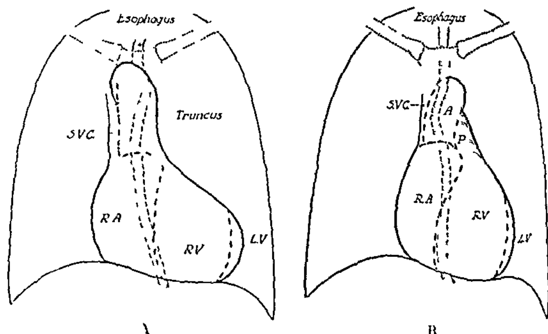


FIGURE XII–14 (A) Truncus arteriosus with a right aortic arch and (B) normal heart Adult

number of vessels given off the transverse portion of the aorta may cause the aortic arch to be fuzzy and indistinct (see Figure XII–20)

In summary, there are five findings of diagnostic importance which should be sought for upon radiological examination

1. Moderate cardiac enlargement with a concave curve at the base of the heart to the left of the sternum
2. A prominent aortic knob which lies at an abnormally high level
3. Absence of the main right and left pulmonary arteries
4. Diffuse fine hilar markings
5. Retro esophageal vessels or bizarre displacement of the esophagus

It is, however, important to remember that retro-esophageal vessels are not always present and that the occurrence of a hilar dance does not exclude the possibility of a 'true' truncus arteriosus with the pulmonary artery arising directly from the aorta

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is subject to considerable variation in most instances the standard leads show a right axis deviation and the precordial leads show evidence of hypertrophy of both ventricles Not infrequently the deflections are of



FIGURE XIV–15 Truncus arteriosus with a left aortic arch Adult

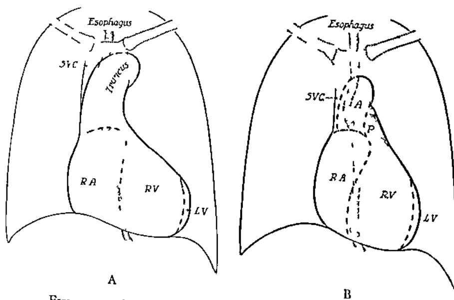


FIGURE XIV–16 (A) Truncus arteriosus with a left aortic arch and (B) normal heart Adult

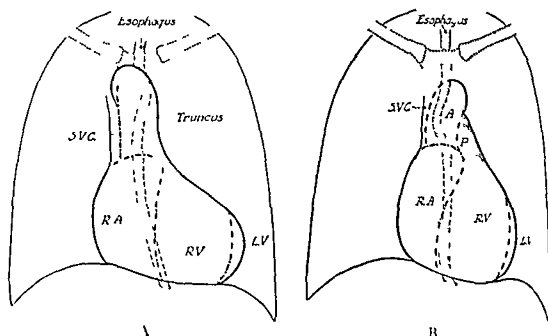


Figure XIV–14 (A) Truncus arteriosus with a right aortic arch and (B) normal heart Adult

number of vessels given off the transverse portion of the aorta may cause the aortic arch to be fuzzy and indistinct (see Figure XIV–20)

In summary, there are five findings of diagnostic importance which should be sought for upon radiological examination

1 Moderate cardiac enlargement with a concave curve at the base of the heart to the left of the sternum
2 A prominent aortic knob which lies at an abnormally high level
3 Absence of the main right and left pulmonary arteries
4 Diffuse fine hilar markings
5 Retro esophageal vessels or bizarre displacement of the esophagus

It is, however, important to remember that retro-esophageal vessels are not always present and that the occurrence of a hilar dance does not exclude the possibility of a 'true' truncus arteriosus with the pulmonary artery arising directly from the aorta

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is subject to considerable variation In most instances the standard leads show a right axis deviation and the precordial leads show evidence of hypertrophy of both ventricles Not infrequently the deflections are of

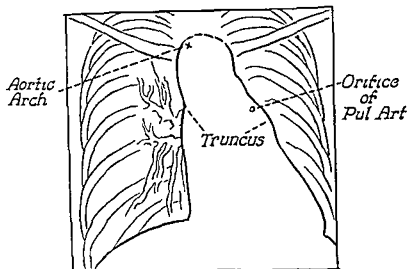


Figure XIV–18 Truncus arteriosus with a single pulmonary artery arising from the base of the truncus (tracing of x ray) (same patient as in Figure XIV–4)



Figure XIV–19 Truncus arteriosus Child

Note the displacement of the esophagus by retro-esophageal vessels



FIGURE XIX-17 Truncus arteriosus with large pulmonary arteries arising from the truncus Child

abnormally great amplitude (see Figure XIX-21) but this finding is not of diagnostic significance Occasionally V_1 shows evidence of right ventricular hypertrophy (see Figure XIX-22), in rare instances the deflection in V_1 is primarily downward (see Figure XIX-23)

SPECIAL TESTS

The circulation time is abnormally short When the pulmonary arteries arise directly from the aorta, the test material may be so rapidly dissipated into the two circulations that a satisfactory end point is not obtainable

The oxygen saturation of the arterial blood varies inversely with the adequacy of the pulmonary blood flow Even though the pulmonary blood flow is adequate or excessive, since blood from both ventricles is directed into the common trunk, there is always slight oxygen unsaturation of the arterial blood The degree of oxygen unsaturation of the arterial blood increases as the circulation

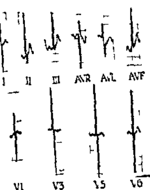


FIGURE XIV-21 Truncus arteriosus (same patient as in Figure XIV-9) Infant

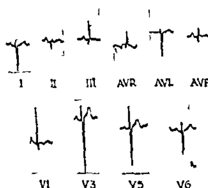


FIGURE XIV-22 Truncus arteriosus Child

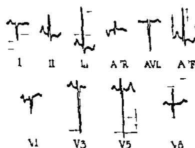


FIGURE XIV-23 Truncus arteriosus Adult

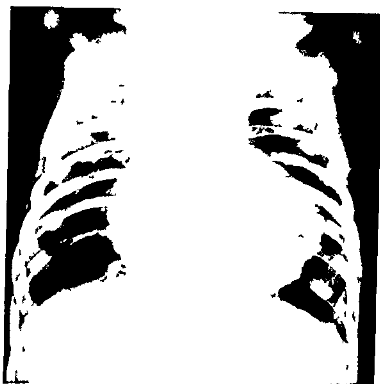
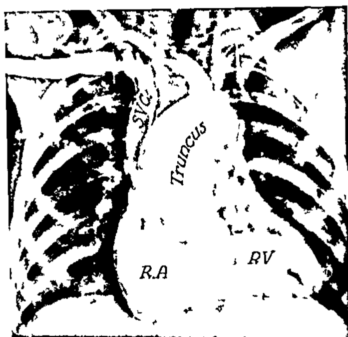


FIGURE XIV–20 Truncus arteriosus and a single ventricle with circulation to the lungs by way of the superior bronchial arteries (same patient as in Figure XIV–6) Infant

to the lungs is reduced When the pulmonary blood flow is meager, the oxygen saturation of the arterial blood may be extremely low Usually, however, the pulmonary blood flow is relatively constant and can be increased by exercise, therefore the oxygen saturation of the arterial blood drops but slightly, if at all, with exercise

The red blood cell count the amount of available hemoglobin and *the hematocrit reading* vary inversely with the adequacy of the pulmonary blood flow Inasmuch as there is always slight oxygen unsaturation of the arterial blood, even when the pulmonary blood flow is normal or excessive, there is always a slight increase in the red blood cell count, the amount of available hemoglobin, and the hematocrit reading Polycythemia increases as the oxygen unsaturation of the arterial blood increases The red blood cell count is frequently between 6 5 and 7 5 million cells per cu mm When the pulmonary blood flow is markedly reduced, the red blood cell count may reach 10 million and there is a proportional increase in the amount of available hemoglobin and in the hematocrit reading

Cardiac catheterization is of no great aid in diagnosis It is usually possible to catheterize the aorta The systolic pressure in the right ventricle is the same



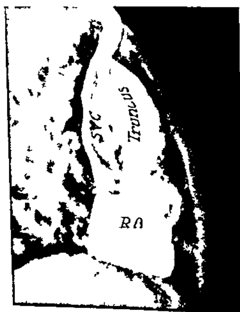


FIGURE XIV–24 Truncus arteriosus with reduced pulmonary blood flow Child

as that in the aorta, and the oxygen content of the blood in the aorta is usually higher than that in the right ventricle It is, however, impossible to catheterize the pulmonary artery directly from the right ventricle

Angiocardiography is of surprisingly little diagnostic aid When the pulmonary arteries arise directly from the truncus, the dye is dissipated so rapidly into the two circulations that frequently it cannot be traced beyond the right ventricle, and yet there is prompt disappearance of the dye from that chamber! When the circulation to the lungs is by way of the bronchial arteries, it is seldom possible to visualize these vessels accurately Angiocardiography is of no aid in the differentiation of this malformation from a tetralogy of Fallot with pulmonary atresia, because in neither instance can the dye enter the pulmonary artery in the normal manner In both instances dye must reach the lungs by way of the collateral circulation An abnormally large aorta," such as is shown in Figure XIV-24, is strongly suggestive of a truncus arteriosus The impression is confirmed by the delineation of a small vessel coursing to the lung

DIAGNOSIS

The occurrence of a continuous murmur over the lungs in a patient with persistent cyanosis and evidence of some cardiac abnormality is almost pathognomonic of a truncus arteriosus Therefore, in the presence of a continuous murmur, the diagnosis of a truncus arteriosus is relatively easy and can be made with a high degree of accuracy

The diagnosis is based upon a combination of findings There may be minimal cyanosis, no limitation of activity, and a loud continuous murmur, there may be moderately severe cyanosis considerable limitation of activity, and a continuous murmur localized over one particular area in the chest, or finally there may be intense cyanosis, marked clubbing and polycythemia, extreme limitation of exercise, and no murmur

The x ray findings are of real aid in diagnosis The heart is slightly to moderately enlarged and there is a concave curve at the base of the heart to the left of the sternum, the 'aorta is abnormally large and arches at an unusually high level, it may arch either to the right or to the left, in the hilar regions no left or right pulmonary artery is visible but numerous fine vascular markings radiate from the hilar regions, delineation of the esophagus with barium may or may not reveal anomalous retro-esophageal vessels The only exception occurs in the rare instance when both pulmonary arteries arise from the left side of the base of the common truncus and there is a conspicuous hilar dance, such patients have a continuous murmur and show little or no cyanosis

tions o cur in combination with another malformation which causes persistent cyanosis, the condition may closely simulate a truncus arteriosus Cardiac catheterization or angiocardiography may be necessary to differentiate the two conditions Either of the tests will show that the pulmonary artery arises from the right ventricle, thus excluding a truncus arteriosus

Rupture of an aneurysm from the sinus of Valsalva into the lesser circulation also causes a continuous murmur The murmur, however, usually develops abruptly in adult life furthermore, the murmur is best heard over the precordium, *not over the lungs* The condition usually leads to progressive cardiac enlargement and cardiac failure

An Eisenmenger complex with aortic insufficiency may be confused with a truncus arteriosus when the diastolic element of the continuous murmur is maximal over the body of the heart The contour of the heart differentiates the two conditions In a truncus arteriosus the shadow at the base of the heart to the left of the sternum is concave, except in the rare instance in which the main pulmonary artery arises from the base of the heart and causes fullness of the pulmonary conus Under the latter circumstances cardiac catheterization may be necessary to differentiate the two malformations

A tetralogy of Fallot with persistent patency of the ductus arteriosus gives essentially the same findings The rarity of such a malformation is shown by the fact that in the author s experience every case in which a continuous murmur was recorded proved at autopsy to be a truncus arteriosus Indeed, among the first 1 000 patients who had a Blalock Taussig operation there was not a single instance of a patient over two years of age with a tetralogy of Fallot in whom the ductus arteriosus had remained patent Therefore, although it is possible to have a tetralogy of Fallot with persistent patency of the ductus arteriosus, the more probable diagnosis is a truncus arteriosus

A tetralogy of Fallot and pulmonary atresia after the obliteration of the ductus arteriosus may require differentiation from a pseudo truncus arteriosus In early infancy the contours of the heart in these two conditions are quite different In a tetralogy of Fallot with pulmonary atresia the heart is essentially normal in shape and is frequently phenomenally small In contrast to this, in a truncus arteriosus the heart is moderately enlarged and the contour is distinctive in that there is no fullness of the pulmonary conus and the ventricle extends abruptly out to the anterior chest wall as a sharp shelf Infants with a tetralogy of Fallot and pulmonary atresia almost always suffer from severe attacks of paroxysmal dyspnea whereas most infants with a truncus arteriosus, although extremely

DIFFERENTIAL DIAGNOSIS

When cyanosis is minimal or absent, a truncus arteriosus may require differentiation from a patent ductus arteriosus or an aortic septal defect, from peripheral pulmonary stenoses, from a rupture of an aneurysm of the sinus of Valsalva or even from an Eisenmenger complex with aortic insufficiency In the presence of moderate cyanosis a truncus arteriosus is usually mistaken for a tetralogy of Fallot with persistent patency of the ductus arteriosus In the presence of intense cyanosis, it may be mistaken for a tetralogy of Fallot with pulmonary atresia, for a non functioning right ventricle, or even for a transposition of the aorta combined with pulmonary atresia

Persistent patency of the ductus arteriosus differs from a truncus arteriosus in that the oxygen saturation of the arterial blood is normal The only exception occurs in early infancy, when the arterial oxygen saturation may be slightly reduced, under such circumstances it usually rises to normal with exercise (see Chapter xx) X ray and fluoroscopy show fullness of the pulmonary conus and conspicuous pulmonary arteries The electrocardiogram usually shows a balanced axis Cardiac catheterization clinches the diagnosis in that the pulmonary artery, not the aorta, is entered from the right ventricle

An aortic septal defect may be confused with a truncus arteriosus, especially when the pulmonary arteries arise from a common orifice on the posterior wall of the truncus arteriosus In a truncus arteriosus this orifice is the point of origin of the pulmonary circulation, whereas in an aortic septal defect the aorta and the pulmonary arteries arise normally from their respective ventricles and there is merely a failure in the completion of the aortic septum The differentiation of these two conditions is vitally important, as surgical closure of an aortic septal defect would restore the heart to normal, whereas closure of the orifice of the pulmonary artery in a truncus arteriosus would cut off all circulation to the lungs and kill the patient

In both conditions there may be a continuous murmur over the base of the heart Cyanosis may be minimal or absent The continuous murmur of an aortic septal defect more closely resembles that of a patent ductus arteriosus than that of a truncus arteriosus It is seldom as loud as that caused by a large ductus arteriosus In doubtful cases, cardiac catheterization may be necessary to determine whether the pulmonary artery or the aorta arises from the right ventricle

Peripheral pulmonary stenoses due to localized constrictions in the branches of the pulmonary arteries may cause a continuous murmur over the lungs which is closely similar to that produced by a truncus arteriosus When such constric

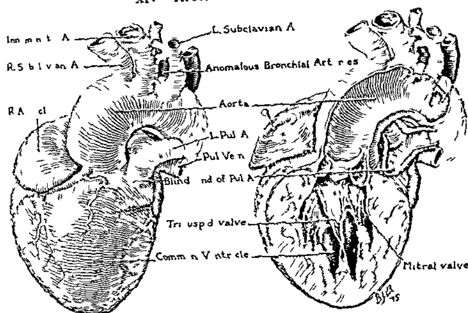


FIGURE XIV-25 Pseudo truncus arteriosus with a single ventricle

tigial pulmonary artery Inasmuch as all the blood returned to the heart is pumped out through a single great vessel, the occurrence of a single ventricle does not fundamentally alter the course of the circulation, as shown in Diagram xiv-3 The basic clinical findings are also unaltered by the absence of the ventricular septum Nevertheless, the occurrence of a single ventricle would be of extreme importance if total correction of the malformation were undertaken

TREATMENT

When the pulmonary arteries arise directly from the common trunk, the danger is that of excessive pulmonary blood flow and pulmonary hypertension In certain instances with excessive pulmonary blood flow and a conspicuous hilar dance, if it were possible to decrease the size of the pulmonary orifice and break the pressure to the lungs, such an operation would benefit the patient Fortunately, in the vast majority of patients the pulmonary vessels which arise from the aorta are smaller than normal and the condition is compatible with a long and active life

If the circulation to the lungs is by way of the small pathways of collateral circulation, the primary difficulty is the reduction of the pulmonary blood flow In such instances a Blalock Taussig operation may be of benefit In order to increase the pulmonary circulation it is not only essential to have a rudimentary

cyanotic at birth, do not suffer from paroxysmal dyspnea and generally do better than other infants with a corresponding degree of cyanosis If a patient with a tetralogy of Fallot and pulmonary atresia survives the closure of the ductus arteriosus, the condition is functionally that of a truncus arteriosus, except that the pulmonary vascular bed is normal because prior to the closure of the ductus arteriosus the pulmonary artery carried blood to the lungs

A non functioning right ventricle causes a square contour of the heart in the anterior posterior position Examination of the heart in the left anterior-oblique position differentiates the two conditions in a non functioning right ventricle, the cardiac shadow does not extend forward anterior to the aorta, whereas in a truncus arteriosus the right ventricle appears definitely enlarged Furthermore, in a non functioning right ventricle there is electrocardiographic evidence of left axis deviation and left ventricular hypertrophy

Complete transposition of the great vessels may occasionally be confused with a truncus arteriosus, because in both conditions there may be persistent cyanosis and relatively pronounced vascular markings Time soon differentiates the two conditions An infant with a complete transposition of the great vessels does poorly, the heart enlarges, and cyanosis increases An infant with a truncus arteriosus and increased flow, if he regains compensation, shows a marked decrease in cyanosis and usually develops a continuous murmur

Transposition of the aorta combined with pulmonary stenosis or atresia is associated with a large aorta, consequently a prominent aortic knob is visible in the anterior posterior position In the left anterior-oblique position the pulmonary window is abnormally clear, in the right anterior oblique position the cardiac shadow causes a sharp shelf like projection from the aorta toward the chest wall The vascular markings extend nearly to the periphery of the lungs when there is a transposition of the great vessels and pulmonary stenosis When the aorta is transposed so far to the left that it occupies the position of the normal pulmonary artery, the unusual course of the aorta and the clear lung fields aid in the differentiation of a transposition of the aorta from a truncus arteriosus (see Chapter X, Section c) The presence or absence of a continuous murmur is also of great diagnostic importance Occasionally the clinical differentiation between the two conditions is extremely difficult

COMMONLY ASSOCIATED MALFORMATIONS

A single ventricle may occur in combination with a truncus arteriosus Figure XIV–25 is a drawing of this type of malformation, in which there was a ves-

DIAGRAM XIV–3

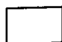
Truncus arteriosus and a single ventricle

In this malformation there is but a single ventricle from which a single great vessel the truncus arteriosus arises The blood is pumped out through the truncus arteriosus to the body and by the pathways of the collateral circulation to the lungs The pulmonary artery if present is a blind tube which has no connection with the truncus arteriosus or with the common ventricle

The blood from the right auricle flows into the *common ventricle and is pumped* out through the truncus arteriosus to the systemic circulation The blood from the body is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle and thence to the common ventricle Inasmuch as the pulmonary artery fails to meet the ventricle and the ductus arteriosus is absent the only way for the blood to reach the lungs is through the bronchial arteries or by some of the other pathways of the collateral circulation The blood which does reach the lungs is returned by the pulmonary veins to the left auricle thence the blood flows into the common ventricle There the cycle starts again

Clinical diagnosis Cyanosis is intense The heart is relatively normal in size but there is no fullness of the pulmonary conus During infancy the contour of the heart resembles that of a non functioning right ventricle As the child grows older the contour of the heart changes and becomes similar to that of a tetralogy of Fallot with an extremely severe pulmonary stenosis The large size of the aortic shadow which lies at an abnormally high level offers a clue to the correct diagnosis If the collateral circulation is by way of the superior bronchial arteries the arch of the aorta may appear fuzzy Unless the cyanosis is extremely intense and the lungs excessively clear, a continuous murmur is usually audible over some areas of the lungs The electrocardiogram generally shows a right axis deviation and evidence of combined hypertrophy

Diagram XIV-3


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

runcus *arteriosus* over rides the ventricular septum, a high ventricular septal defect *is an integral part of the malformation* Furthermore, since the truncus arteriosus receives blood from both ventricles, there is always some degree of oxygen unsaturation of the arterial blood

The clinical findings vary with the adequacy of the pulmonary blood flow If the pulmonary blood flow is adequate, cyanosis is minimal, polycythemia and oxygen unsaturation are proportionately *slight, and the patient may enjoy a rela tively normal life* If *the circulation to the lungs is established through enlarged bronchial* arteries, cyanosis is moderately intense and the red blood cell count is slightly to moderately elevated, under such circumstances the patient s capacity for exercise is moderately *reduced* Nevertheless, the condition is compatible with life for a number of years If, however, the circulation to the lungs is by way of small bronchial arteries or other minute vessels, cyanosis and polycythemia are intense, the patient s exercise tolerance is *extremely limited*

The heart is usually slightly to moderately enlarged, the second sound at the base *is accentuated* but never reduplicated The murmurs vary with the ade quacy of the pulmonary blood flow When the pulmonary blood flow is ade quate or excessive, there is a loud continuous murmur, when it is moderately reduced, the continuous murmur is usually localized to some specific portion of the chest when the pulmonary blood flow is minimal, there may be no murmur

X ray and fluoroscopy show slight to moderate cardiac enlargement, a large aorta, a high aortic arch, *absence of fullness* of the pulmonary conus, and absence of the normal shadows cast by the pulmonary arteries Usually the lung fields are clear and numerous fine shadows are seen to radiate from the hilar regions The esophagram may show evidence of retro-esophageal vessels

The electrocardiogram usually shows right axis deviation in the standard leads the precordial leads are variable but generally show evidence of hyper trophy of both ventricles

The circulation time is abnormally short

Cardiac catheterization and angiocardiography are of diagnostic aid mainly in the exclusion of other conditions

The *diagnosis is based upon the finding* of a continuous murmur over the lungs in a patient with persistent cyanosis, combined with characteristic x ray and fluoroscopic findings

In the absence of cyanosis a truncus arteriosus *may require* differentiation from a patent ductus arteriosus or an aortic septal defect, from *peripheral pul* monary stenoses from a rupture of an aneurysm of the sinus of Valsalva, or from an Eisenmenger complex with aortic insufficiency In the presence of cyanosis,

pulmonary artery of sufficient size to carry blood to the lungs, but also essential to have a normal pulmonary vascular bed. If such is the case, since both auricles are normally formed and both ventricles pump the blood into the common trunk, the creation of an artificial ductus arteriosus increases the pulmonary blood flow without any significant alteration in the work required of the heart. Under such circumstances a Blalock-Taussig operation is of great benefit.

Unfortunately, in many instances of a pseudo truncus arteriosus, even though the pulmonary artery is of fair size, the pulmonary vascular bed appears unable to open up in the normal manner and carry the increased volume of blood to the lungs. When such is the situation, thromboses develop distal to the anastomosis. Not only does the patient fail to derive benefit from the operation but generally he does not survive. The over all mortality rate for patients with truncus arteriosus is about 25 per cent. Therefore, if the child is only slightly incapacitated, operation is contraindicated.

PROGNOSIS

The prognosis depends upon the pulmonary blood flow. If large pulmonary arteries arise directly from the common trunk, the infant may die of excessive pulmonary blood flow within the first few days.

If the pulmonary artery arises from the truncus arteriosus at a slightly higher level or from a common orifice, the condition may be compatible with relative longevity.

Indeed, if the collateral circulation is such that there is a relatively adequate pulmonary blood flow, the prognosis is reasonably good. Even though the patient is cyanotic, there is only moderate polycythemia, his exercise tolerance will be good and for many years he may enjoy a relatively normal life.

If the pulmonary blood flow is markedly reduced, a Blalock Taussig operation is indicated. If this is successful, the prognosis will be greatly improved. Without operation, the prognosis is poor.

SUMMARY

A truncus arteriosus is the result of an early arrest in the development of the great vessels, either the pulmonary artery arises directly from the base of the common trunk, or a vestigial pulmonary artery fails to reach the aortic trunk and the circulation to the lungs is by way of the bronchial arteries. In both instances the blood from the two ventricles is pumped out into a common vessel which directs the blood to the body and to the lungs. Both ventricles pump against systemic pressure, both ventricles are thick walled. Inasmuch as the

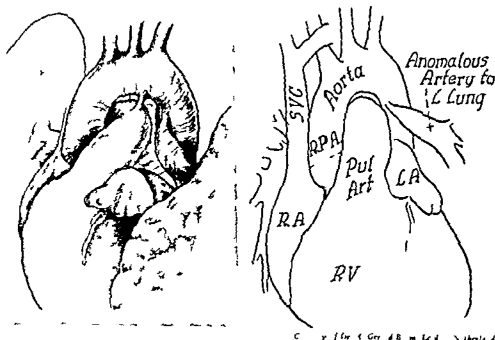


FIGURE XIV-26 Hemi truncus arteriosus (same patient as in Figures XIV-28 29) Child

COURSE OF THE CIRCULATION

During fetal life although the single pulmonary artery goes to only one lung, so little blood flows through the pulmonary artery that the absence of the other pulmonary artery places no strain on the fetal circulation At birth the heart is normal in size (see Figure XIV-27)

After birth the blood from the right auricle flows into the right ventricle and thence is pumped out into the main pulmonary artery and through the single pulmonary vessel to *one* lung, where it is oxygenated, and returned in the normal manner to the left auricle The blood from the left auricle flows into the left ventricle and is pumped out through the aorta to the systemic circulation and also to the lung which receives its blood from the anomalous vessel that arises from the aorta This lung receives fully oxygenated blood, circulates through the lung, and is then returned to the left auricle The blood which circulates through the body is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again (see Diagram XIV-4)

it is frequently mistaken for a tetralogy of Fallot with persistent patency of the ductus arteriosus When cyanosis is intense, a truncus arteriosus is anatomically and functionally closely related to that which occurs in a patient with a tetralogy of Fallot combined with pulmonary atresia who survives the closure of the ductus arteriosus Nevertheless, the difference between these two conditions is important because of the difference in the operative risk Occasionally the condition requires differentiation from a complete transposition of the great vessels with or without pulmonary atresia or from a non functioning right ventricle

A patient with severe reduction in the pulmonary blood flow may be greatly benefited by a Blalock-Taussig operation, provided not only that a vestigial pulmonary artery exists, but also that the pulmonary vascular bed is normal and the pulmonary artery is able to direct blood to the lungs in the normal manner The mortality rate from operation is approximately 25 per cent

The prognosis varies with the adequacy of the pulmonary blood flow If the pulmonary blood flow is relatively adequate, the prognosis is excellent When the pulmonary blood flow is severely reduced, the prognosis may be greatly improved by successful surgery

B *Hemi Truncus Arteriosus*

In a hemi truncus arteriosus, one pulmonary artery arises directly from the aorta, as it does in a truncus arteriosus, and the other pulmonary artery arises normally from the right ventricle Although a single pulmonary artery occurring in combination with various malformations, such as a tetralogy of Fallot, may be considered as a variant of a hemi truncus arteriosus, the following discussion is concerned with the condition when it occurs as an isolated abnormality

NATURE OF THE MALFORMATION

"Hemi truncus arteriosus" is the name given to the condition in which the heart itself is normal and the great vessels arise normally but the main pulmonary artery supplies only one lung and the other lung is supplied by a vessel which arises directly from the aorta The single pulmonary artery may go to either the right or the left lung The author has studied one case in which the pulmonary artery went to the left lung and Dr A Gerard Brom told her of a case in which the pulmonary artery was directed to the right lung (see Figure XIV–26) In both instances the circulation to the other lung was by way of an anomalous vessel which arose from the aorta

CARDIAC FINDINGS

The heart is normal in size and shape The *heart sounds* are normal There is, however, a *continuous murmur* audible over the lung which is supplied from the aorta When the murmur is heard on the left side, it may be confused with that of persistent patency of the ductus arteriosus The murmur has a more humming quality and is usually more widely transmitted than that of a patent ductus arteriosus

X RAY FINDINGS

The x ray findings are not remarkable (see Figure xiv-28)

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is usually normal

SPECIAL TESTS

Cardiac catheterization shows that the circulation is normal Indeed, when patency of the ductus arteriosus is suspected, the absence of any increase in the oxygen content of the blood in the pulmonary artery gives the clue to the diagnosis of a hemi truncus arteriosus

Angiocardiography is diagnostic in that one pulmonary artery is visualized with the right side of the heart and the other with the left The pulmonary artery which arises from the right ventricle fills normally (see Figure xiv-29) but no dye enters the other lung until the aorta is visualized and then only if the vessel is sufficiently large to permit opacification of the pulmonary vascular bed

DIAGNOSIS

A hemi truncus arteriosus may be suspected when a continuous murmur is well heard over the right lung, especially in an asymptomatic patient with a normal left aortic arch When the continuous murmur is heard over the left side of the chest it may easily be confused with that of patency of the ductus arteriosus

DIFFERENTIAL DIAGNOSIS

The condition may closely simulate patency of the ductus arteriosus, an aortic septal defect or peripheral stenoses of the pulmonary artery All these conditions are readily differentiated by angiocardiography or by cardiac catheterization A hemi truncus arteriosus, however, is so rare that neither of these

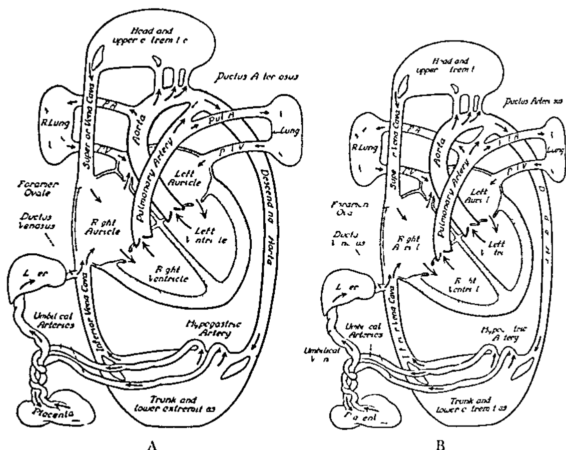


Figure XII-27 Fetal circulation (A) Hemi truncus arteriosus and (B) normal heart

PHYSIOLOGY OF THE MALFORMATION

The entire oxygenation of the blood occurs in the lung which receives venous blood from the pulmonary artery The lung which receives blood from the aorta is non functioning as far as oxygenation is concerned, but does aid in the normal expansion of the chest and prevents the displacement of the mediastinum The outstanding physiological change caused by this anomaly is the difference in the amount of oxygen taken up and the amount of carbon dioxide given off in the two lungs Aside from this abnormality, the basic hemodynamics are normal

CLINICAL FINDINGS

The patient is generally asymptomatic The condition is usually detected on a routine physical examination

Cyanosis is absent There is no admixture of venous and arterial blood, the oxygen saturation of the arterial blood is normal

Diagram XIV–4

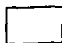
Hemi truncus arteriosus

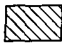
The essential feature of this malformation is that there is but a single pulmonary artery which goes to only one lung the other lung receives its blood from a vessel which arises from the aorta

The blood from the right auricle flows into the right ventricle and is pumped out into the pulmonary artery, thence it flows to one lung only In that lung the blood is oxygenated in the normal manner and is returned to the left auricle The blood from the left auricle flows into the left ventricle and is pumped out through the aorta to the systemic circulation and through an anomalous artery (A A) to the other lung The fully oxygenated blood thus directed to the lungs through the anomalous vessel passes through the capillaries and is returned to the left auricle It follows that all the blood which the left auricle receives is fully oxygenated this blood flows into the left ventricle and out into the aorta The major part of the blood in the aorta is directed to the systemic circulation and the venous blood from the body is returned by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

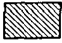
Clinical diagnosis The patient is asymptomatic and shows no cyanosis The heart is normal in size and shape A continuous murmur is audible over the lung on the side which has the anomalous circulation The electrocardiogram is normal

DIAGRAM XIV–4


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood



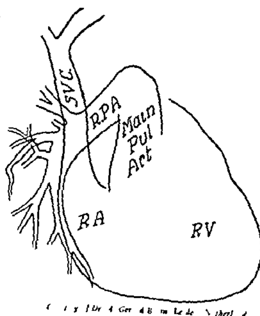


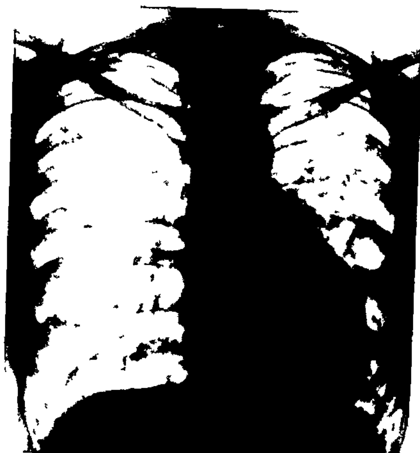
FIGURE XII–29 Hemi truncus arteriosus (same patient as in Figure XII–26) Child

Chapter XXI) Although the patient is living on one lung and the other lung is of no use in the oxygenation of the blood, there is no evidence that the functionless lung is prone to infection Actually that lung aids in the normal growth and expansion of the chest Hence pneumonectomy is contraindicated

SUMMARY

A hemi truncus arteriosus means that the circulation to one lung is similar to that of a truncus arteriosus and the circulation to the other lung is normal Thus a single pulmonary artery supplies one lung and an anomalous vessel which arises from the aorta directs the blood to the other lung Although the entire oxygenation of the blood occurs in the lung which has a normal supply, the patient is asymptomatic The existence of an abnormality is readily detected upon physical examination, as there is a continuous murmur over the lung which receives its blood from the aorta When this murmur is heard on the left side, the condition may be mistaken for patency of the ductus arteriosus

The condition requires differentiation from persistent patency of the ductus arteriosus and from peripheral pulmonary stenoses Both these conditions can readily be differentiated from a hemi truncus arteriosus by angiocardiography When there is but a single pulmonary artery the dye passes from the right ven



Courtesy of Dr. Gerard Brom, Leiden, Netherlands

FIGURE XIV-28 Hemi truncus arteriosus (same patient as in Figure XIV-26) Child

procedures is indicated as a routine to exclude the possibility in a patient with classic signs of patency of the ductus arteriosus An occasional exploratory thoracotomy carries no greater risk than would a routine angiocardiogram performed on every patient suspected of patency of the ductus arteriosus

Peripheral pulmonary stenoses are usually associated with hypertension in the proximal portion of the pulmonary artery This causes accentuation of the second sound at the base of the heart to the left of the sternum and, furthermore, the electrocardiogram usually shows a right axis deviation and evidence of right ventricular hypertrophy

TREATMENT

There is none, and none is necessary If operation is performed for patency of the ductus arteriosus and none is found, the chest should be closed unless further dissection is deemed advisable to rule out an aortic septal defect (see

CHAPTER XV

A SINGLE VENTRICLE AND A RUDIMENTARY OUTLET CHAMBER

A SINGLE ventricle is the result of an extremely early arrest in the development of the heart It produces an entirely different architecture of the ventricles from that found in any other malformation [1]

Furthermore, the failure of the formation of the interventricular septum is frequently associated with variations in the size and position of the great vessels Either great vessel may be large, either may be small In addition there may be a transposition of the great vessels Consequently there is great variation in the clinical picture The diagnosis is correspondingly difficult and is frequently made only by the exclusion of other conditions Nevertheless, it is an important malformation

A single ventricle is relatively common when the primitive cardiac loop swings to the right instead of developing in the normal manner with the apex of the heart pointing to the left Hence it is common in patients with dextrorotation of the heart A single ventricle, however, may occur when the heart occupies its normal position

A brief review of the embryology will help to clarify its nature.

EMBRYOLOGY

When the primitive cardiac tube bulges forward and swings to the right, the anterior portion of it becomes the bulbus cordis and the posterior part of the loop forms the common ventricle A single great vessel arises from the bulbus cordis Normally, as the heart develops, the ridge which separates the bulbus cordis from the common ventricle atrophies Coincidentally with the disappearance of this ridge the aortic septum develops and divides the aortic trunk into the aorta and the pulmonary artery At the same time the development of the ventricular septum divides the common ventricle into two chambers The posterior portion of the common ventricle becomes the left ventricle, from which the aorta arises, the lower portion of the bulbus cordis expands and fuses with the anterior portion of the common ventricle to form the right ventricle, from which the pulmonary artery arises

In the malformation under discussion the ventricular septum fails to develop

tricle to one lung only, and no dye reaches the other lung until after the aorta has been visualized

A hemi truncus arteriosus causes no difficulty and requires no treatment In deed, pneumonectomy is contraindicated

References

1 Humphreys, E M Truncus arteriosus communis persistens criteria for identification of common arterial trunk, with report of a case with 4 semilunar cusps Arch Path. 14 671–700, 1932

2 Christeller, E Funktionelles und Anatomisches bei der angeborenen Verengerung und dem angeborenen Verschluss der Lungenarterie, insbesondere über die arteriellen Kol lateralbahnen bei diesen Zustanden Virchows Arch f path Anat 223 40–57 1916–17

3 White, P D Personal communication

4 Collister, R M Collateral Circulation in Stenosis of the Great Vessels Leiden, Uit geverij Pompe, 1952

5 Collister, R M J Dankmeijer H A Snellen and W H van der Wel Rontgenonder zoek van Hartmisvormingen Ned Tijdschrift v Geneeskunde J vol 95 part 3 no 34 p 2504 1951

6 MacGilpin H H Jr Truncus arteriosus communis persistens Am Heart J 39 615–625, 1950

7 Timm C Die Blutbewegung in der Aorta Rontgenkinematographische Untersuchun gen mit Zeitdehnung an Katzen und Hunden Arch f d ges Physiol 249 261–279 1947

8 Reynolds S R M F W Light Jr G M Ardran and M M L Prichard The quali tative nature of the pulsatile flow in the umbilical blood vessels with observations on the flow in the aorta Bull Johns Hopkins Hosp 91 83–104 1952

9 Danelius G Absence of the hilar shadow a diagnostic sign in rare congenital cardiac malformations (truncus arteriosus solitarius with heterotopic pulmonary blood supply) Am J Roentgenol 47 870–876 1942

casionally both vessels may arise from the diminutive chamber, more often there is a transposition of the great vessels Under the latter circumstances the aorta arises from the diminutive chamber and the pulmonary artery from the common ventricle Ordinarily the vessel which arises from the rudimentary chamber is diminutive in size and that which arises from the common ventricle is of normal size or enlarged It is, however, theoretically possible for the pulmonary artery to be stenotic when it arises from the common ventricle

The malformation is further complicated when a transposition of the great vessels is combined with the anomalous insertion of the *chordae tendineae* of the tricuspid valve along the base of the rudimentary outlet chamber in such a manner that the blood from the right auricle is directed primarily into the rudimentary chamber and hence to the transposed aorta Figure xv-2 shows a drawing of such a specimen and Figure xv-3 shows the manner in which the blood is directed from the two auricles into the common ventricle and out into the aorta and the pulmonary artery This combination of anomalies, which was reported by Lambert is termed the Lambert heart The malformation is not rare, it produces a distinctive clinical picture

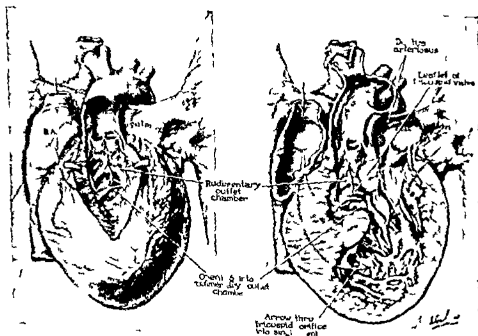


FIGURE xv-2 Single ventricle (Lambert heart) (same patient as in Figures xv-3 8,9) Child

and the bulbus cordis persists as a rudimentary outlet chamber. The arrest in the development of the heart may occur so early that both great vessels arise from the persistent bulbus cordis, or one may arise from the common ventricle and one from the rudimentary outlet chamber.

NATURE OF THE MALFORMATION

The essential feature of the malformation is a single ventricle with a rudimentary outlet chamber which lies in the region normally occupied by the outflow tract of the right ventricle. The diminutive chamber is separated from the main ventricle by a muscular ridge (see Figure xv-1). No valve or membrane guards its orifice. There is free communication between the common ventricle, which receives the blood from both auricles, and the small chamber from which one or both the great vessels arise. If the great vessels develop normally, the aorta arises from the common ventricle and the pulmonary artery from the rudimentary chamber. It is, however, common to find that the arrest in the development of the ventricle is associated with an abnormality of the great vessels. Of

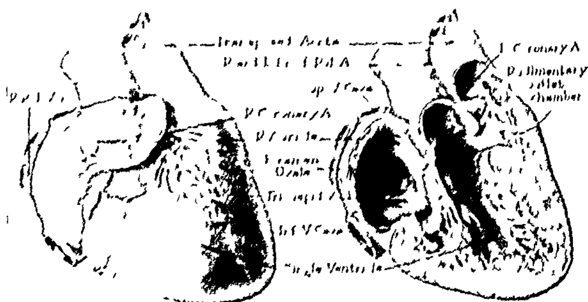


FIGURE xv-1. Single ventricle and a rudimentary outlet chamber from which the transposed aorta arises (same patient as in Figure x-7). Infant.

The foramen ovale is incompletely covered by a valve. In this case the mitral valve was atretic. Usually it, too, opens into the common ventricle.

casionally both vessels may arise from the diminutive chamber, more often there is a transposition of the great vessels Under the latter circumstances the aorta arises from the diminutive chamber and the pulmonary artery from the common ventricle Ordinarily the vessel which arises from the rudimentary chamber is diminutive in size and that which arises from the common ventricle is of normal size or enlarged It is, however, theoretically possible for the pulmonary artery to be stenotic when it arises from the common ventricle

The malformation is further complicated when a transposition of the great vessels is combined with the anomalous insertion of the chordae tendineae of the tricuspid valve along the base of the rudimentary outlet chamber in such a manner that the blood from the right auricle is directed primarily into the rudimentary chamber and hence to the transposed aorta Figure XV–2 shows a drawing of such a specimen and Figure XV–3 shows the manner in which the blood is directed from the two auricles into the common ventricle and out into the aorta and the pulmonary artery This combination of anomalies, which was reported by Lambert is termed the *Lambert heart* The malformation is not rare, it produces a distinctive clinical picture

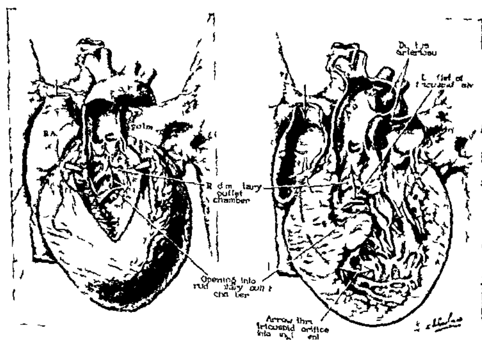


FIGURE XV–2 Single ventricle (Lambert heart) (same patient as in Figures XV–3 8 9) Child

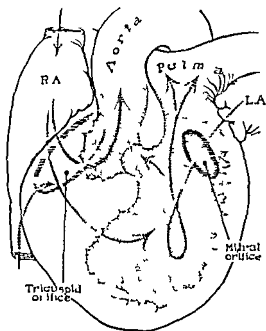


FIGURE xi-3 Single ventricle (Lambert heart) (same patient as in Figure xi-2) Child

The arrest in the development of the ventricles may or may not be associated with a malformation of the auricular septum If the auricular septum develops normally, the heart is a triloculate one or *cor triloculare biatriatum* (three-cham bered heart with one ventricle and two auricles) Under such circumstances the mitral and tricuspid valves open into the common ventricle If, on the other hand, there is an arrest in the development of the auricular septum so that it, too, fails to form, the resultant malformation is a *cor biloculare,* or a biloculate heart *with a single auricle, a common atrioventricular valve, and a single ven tricle*

COURSE OF THE CIRCULATION

During fetal life, although the structure of the heart is extremely primitive, it suffices to meet the needs of the fetus Inasmuch as the lungs are of no func tional importance, it makes little difference which of the great vessels is given off from the diminutive chamber and which from the common ventricle At birth the heart is normal in size

After birth the fact that one of the great vessels is diminutive in size has a profound effect upon the circulation The blood from the right auricle flows through *the tricuspid valve into the common ventricle and the blood from the* left auricle flows through the mitral valve into the common ventricle Therefore, regardless of the structure of the auricular septum, there is complete admixture of the venous and arterial blood in the common ventricle As the ventricle fills, blood also flows into the rudimentary chamber Consequently the same admix

ture of venous and arterial blood is pumped out into both great vessels If both great vessels arise from the diminutive chamber and are approximately the same size, approximately the same volume of blood is pumped out through the pulmonary artery to the lungs as is pumped out through the aorta to the systemic circulation The blood from the lungs is returned by the pulmonary veins to the left auricle and the blood from the body is returned by the superior and inferior venae cavae to the right auricle The blood from both auricles again flows into the common ventricle Inasmuch as the pulmonary blood flow is approximately equal to the systemic blood flow, there may be no visible cyanosis Frequently, however, a relatively small volume of blood reaches the lungs for oxygenation and the systemic circulation is also meager, therefore the infant shows persistent cyanosis (see Diagram xv-1)

As a rule, one great vessel is given off the main ventricle and the other off the rudimentary chamber, and furthermore the vessel which is given off the main chamber is usually the larger of the two Consequently, although the course of the circulation is fundamentally the same, owing to the difference in the size of the two vessels, the resultant admixture of venous and arterial blood is quite different

When the pulmonary artery arises from the main ventricle and the aorta from the rudimentary chamber, a large volume of blood is directed to the lungs and a relatively small amount is directed to the body Hence a large amount of *fully* oxygenated blood is returned to the left auricle, whereas a relatively small amount of venous blood is returned to the right auricle Consequently a large volume of oxygenated blood is mixed with a small amount of venous blood and the resultant admixture is above the threshold of visible cyanosis (see Diagram xv-2) Cyanosis is absent

When the aorta arises from the common ventricle and the pulmonary artery from the diminutive chamber, the situation is quite different Under such circumstances the pulmonary artery may be diminutive or the pulmonary orifice may be stenotic Thus the body receives an *adequate or excessive* volume of blood but only a small volume of blood goes to the lungs Hence a small volume of oxygenated blood is returned to the left auricle *to mix* with a large volume of venous blood returned from the body by the superior and inferior venae cavae to the right auricle Hence cyanosis is intense (see Diagram xv-3)

When a transposition of the great vessels is combined with the anomalous insertion of the chordae tendineae of the tricuspid valve into the base of the rudimentary chamber the situation is still further complicated Under such circum

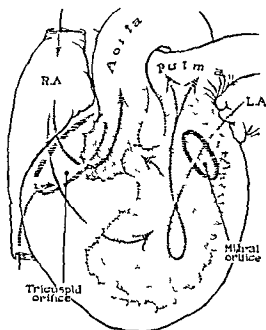


Figure XX-3 Single ventricle (Lambert heart) (same patient as in Figure XX-2) Child

The arrest in the development of the ventricles may or may not be associated with a malformation of the auricular septum If the auricular septum develops normally, the heart is a triloculate one or *cor triloculare biatriatum* (three-chambered heart with one ventricle and two auricles) Under such circumstances the mitral and tricuspid valves open into the common ventricle If, on the other hand, there is an arrest in the development of the auricular septum so that it, too, fails to form, the resultant malformation is a *cor biloculare* or a biloculate heart with a single auricle, a common atrioventricular valve, and a single ventricle

COURSE OF THE CIRCULATION

During fetal life, although the structure of the heart is extremely primitive, it suffices to meet the needs of the fetus Inasmuch as the lungs are of no functional importance, it makes little difference which of the great vessels is given off from the diminutive chamber and which from the common ventricle At birth the heart is normal in size

After birth the fact that one of the great vessels is diminutive in size has a profound effect upon the circulation The blood from the right auricle flows through the tricuspid valve into the common ventricle and the blood from the left auricle flows through the mitral valve into the common ventricle Therefore, regardless of the structure of the auricular septum, there is complete admixture of the venous and arterial blood in the common ventricle As the ventricle fills, blood also flows into the rudimentary chamber Consequently the same admix

DIAGRAM XV-1

Single auricle and a single ventricle with a rudimentary outlet chamber from which both great vessels arise

In this malformation the auricular septum is so rudimentary that there is but a single large auricle Both the tricuspid valve and the mitral valve may be present or there may be a common atrioventricular valve which opens into the ventricle *There is but* a single ventricle with a rudimentary outlet chamber from which both the pulmonary artery and the aorta arise The great vessels are diminutive in size

The blood from the common auricle flows into the common ventricle Inasmuch as both great vessels arise from the rudimentary outlet chamber, all the blood from the common ventricle must be forced from this small chamber into the aorta and the pulmonary artery Since both great vessels arise from the same chamber, the pressure in the aorta and the pulmonary artery is the same The blood which is pumped into the aorta goes to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right side of the auricle. The blood which is pumped into the pulmonary artery goes to the lungs, where it is oxygenated, and is returned by the pulmonary veins to the left side of the auricle There the cycle starts again

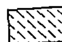
Clinical diagnosis is based upon the contour of the heart The size of the heart is not greatly altered but the rudimentary outlet chamber causes a prominence of the pulmonary conus in the anterior posterior position in the left anterior-oblique position however there is no enlargement of the right ventricle The increased pressure in the ventricle is transmitted to the auricle and this, in turn, causes dilatation of the superior vena cava

There is complete admixture of the venous and arterial blood in both the common auricle and the single ventricle Furthermore inasmuch as the two great vessels are of equal size approximately equal volumes of blood are directed to the pulmonary and systemic circulations there is persistent cyanosis The mechanism for pumping the blood from the single ventricle through the rudimentary outlet chamber is inefficient The condition is rarely compatible with life for more than a few months

Diagram XV-1

Head and upper extremities
R Lung
L Lung
S.V.C.
P.A.
Aorta
Pul A
Pulmonary A
PV
Pul V
L A
R.A.
Single Ventricle
Descending Aorta
Rudimentary Outlet Chamber
I.V.C.
Trunk and lower extremities


Arterial blood (fully saturated)


Small admixture of venous blood
No visible cyanosis


Venous and arterial blood
Cyanosis visible


Venous blood

Diagram XV-1

Single auricle and a single ventricle with a rudimentary outlet chamber from which both great vessels arise

In this malformation the auricular septum is so rudimentary that there is but a single large auricle Both the tricuspid valve and the mitral valve may be present or there may be a common atrioventricular valve which opens into the ventricle. There is but a single ventricle with a rudimentary outlet chamber from which both the pulmonary artery and the aorta arise. The great vessels are diminutive in size

The blood from the common auricle flows into the common ventricle Inasmuch as both great vessels arise from the rudimentary outlet chamber, all the blood from the common ventricle must be forced from this small chamber into the aorta and the pulmonary artery Since both great vessels arise from the same chamber *the pressure* in the aorta and the pulmonary artery is the same The blood which is pumped into the aorta goes to the systemic circulation and is returned by the superior vena cava and the inferior vena cava to the right side of the auricle The blood which is pumped into the pulmonary artery goes to the lungs where it is oxygenated and is returned by the pulmonary veins to the left side of the auricle. There the cycle starts again

Clinical diagnosis is based upon the contour of the heart The size of the heart is not greatly altered but the rudimentary outlet chamber causes a prominence of the pulmonary conus in the anterior posterior position in the left anterior-oblique position however there is no enlargement of the right ventricle The increased pressure in the ventricle is transmitted to the auricle and this in turn causes dilatation of the superior vena cava

There is complete admixture of the venous and arterial blood in both the common auricle and the single ventricle. Furthermore, inasmuch as the two great vessels are of equal size approximately equal volumes of blood are directed to the pulmonary and systemic circulations there is persistent cyanosis The mechanism for pumping the blood from the single ventricle through the rudimentary outlet chamber is inefficient The condition is rarely compatible with life for more than a few months

Diagram xv–2

Head and upper extremities
R Lung
P.A.
S.V.C.
Aorta
Pul A
L Lung
PV
Pul V
Pulmonary Artery
L A
R.A.
Descending Aorta
Single Ventricle
Rudimentary Outlet Chamber
I.V.C.
Trunk and lower extremities



Arterial blood (fully saturated)



Small admixture of venous blood
No visible cyanosis



Venous and arterial blood
Cyanosis visible



Venous blood

Diagram XV-2

Single ventricle with a rudimentary outlet chamber from which the aorta arises

In this malformation there are two auricles and a single ventricle with a rudimentary outlet chamber The great vessels are transposed The pulmonary artery arises from the main ventricle and is of normal size the aorta arises from the diminutive outlet chamber and is of small caliber

The blood from both auricles flows into the common ventricle As the common ventricle fills the blood flows into the rudimentary outlet chamber The blood from the common ventricle is pumped into the pulmonary artery and that from the rudimentary chamber into the aorta The blood in the pulmonary artery flows to the lungs and the oxygenated blood is returned by the pulmonary veins to the left auricle The blood in the aorta flows into the *systemic circulation* and is returned in the normal fashion by the superior and inferior venae cavae to the right auricle Regardless of whether or not the auricular septum is sufficiently well developed to prevent an appreciable admixture of the blood in the two auricles, there is complete admixture of venous and arterial blood in the common ventricle. Inasmuch as the pulmonary artery is larger than the aorta and is given off the common ventricle, a far greater volume of blood flows to the lungs than to the systemic circulation Furthermore, the blood *is* pumped to the lungs under systemic pressure, hence there is severe pulmonary hypertension Nevertheless the volume of oxygenated blood returned to the left auricle is larger than the volume of venous blood returned to the right auricle. Usually the volume of oxygenated blood returned from the lungs is sufficiently great so that there is no visible cyanosis

Clinical diagnosis is based upon the contour of the heart. In infancy, in the anterior posterior position the rudimentary outlet chamber causes a prominence in the region of the pulmonary conus In older children this chamber is no longer visible and the aortic shadow is narrow In both instances, in the left anterior-oblique position, there is no enlargement of the right ventricle. In addition, there is usually great exaggeration of the hilar shadows due to the large volume of blood flowing through the pulmonary circulation Clubbing and cyanosis are absent Murmurs may or may not be present and are of *no* diagnostic aid The electrocardiogram is variable but may offer a clue to the diagnosis in that there is often a discrepancy between the axis deviation in the standard leads and the evidence of ventricular hypertrophy in the unipolar precordial leads

DIAGRAM XV–3

Head and upper extremities
R Lung
S V C
P A
P V
Aorta
Pul A
L Lung
Pul V
Pulmonary Art
L A
R A
Single Ventricle
Descending Aorta
Rudimentary Outlet Chamber
I V C
Trunk and lower extremities

Arterial blood (fully saturated)

Venous and arterial blood
Cyanosis visible

Small admixture of venous blood
No visible cyanosis

Venous blood

Diagram XV-3

Single ventricle with a rudimentary outlet chamber from which the pulmonary artery arises

This malformation represents an early arrest in the development of the heart There is but a single ventricle with a rudimentary outlet chamber The aorta lies more posteriorly than does the pulmonary artery It is given off the common ventricle and is of normal size, whereas the pulmonary artery is given off the rudimentary chamber and is of small caliber

The blood from the right auricle together with that from the left auricle passes through the atrioventricular valve into the common ventricle as the common ventricle fills blood also flows into the rudimentary outlet chamber The blood in the common ventricle is pumped into the aorta and that in the rudimentary outlet chamber is pumped into the pulmonary artery The blood in the aorta flows into the systemic circulation and is returned in the normal fashion by the superior and inferior venae cavae to the right auricle The blood in the pulmonary artery flows into the lungs and is returned by the pulmonary veins to the left auricle There the cycle starts again

Inasmuch as the aorta is large and arises from the common ventricle and the pulmonary artery is small and receives only the blood from the rudimentary chamber a much greater volume of blood goes to the systemic circulation than goes to the lungs for oxygenation Hence the volume of venous blood returned to the right auricle is larger than the volume of oxygenated blood returned to the left auricle Thus in the common ventricle there is admixture of a large volume of venous blood with a small volume of oxygenated blood Cyanosis is intense.

Clinical diagnosis is based upon the finding of persistent cyanosis and a heart with a contour which is essentially the same as that in other cases of a single ventricle with a rudimentary outlet chamber In infancy the heart is *not enlarged*, *nevertheless* in the anterior posterior position there is prominence of the pulmonary conus, but in the left anterior-oblique position there is no enlargement of the right ventricle In older children the prominence of the pulmonary conus disappears and the contour of the heart resembles that of an extreme tetralogy of Fallot Cyanosis is intense The condition is usually compatible with life for only a few months

stances the blood from the right auricle is directed mainly to the rudimentary outlet chamber and thence directed into the transposed aorta The blood from the left auricle is directed into the common ventricle Consequently even though the pulmonary artery may be greatly enlarged, the systemic circulation receives mainly venous blood The patient will be intensely cyanotic The course of the circulation is shown in Diagram xv–4

PHYSIOLOGY OF THE MALFORMATION

The two features of the common ventricle which affect the physiology of the malformation are the free admixture of venous and arterial blood in the common ventricle and the common ejectile force with which the blood from the ventricle is pumped into both great vessels Thus there is always some oxygen unsaturation of the arterial blood and, except when there is pulmonary stenosis, there is always pulmonary hypertension The high pressure with which the blood is ejected into the lungs eventually leads to intimal proliferation which causes progressive narrowing of the pulmonary vascular bed and thereby increases the pulmonary resistance Thus over a period of years less blood is pumped through the pulmonary circulation and more blood is directed into the aorta This means that the volume of blood which reaches the lungs for oxygenation is decreased, hence a smaller volume of oxygenated blood is returned to the left auricle and the oxygen content of the blood in the common ventricle is proportionally reduced Consequently, although a progressively greater volume of blood is directed to the systemic circulation, the oxygen saturation of the arterial blood is gradually reduced As the oxygen unsaturation slowly increases, cyanosis, even if not initially obvious, sooner or later becomes apparent and gradually deepens These changes, however, take years to develop Needless to say, in the presence of pulmonary stenosis, pulmonary hypertension never develops

CLINICAL FINDINGS

Inasmuch as a single ventricle may be associated with decreased or increased pulmonary blood flow, and furthermore the increased pulmonary blood flow may be associated with an anomalous insertion of the tricuspid valve of such a nature as to direct venous blood to the aorta, the clinical manifestations are protean

The history is often significant Infants who show no cyanosis frequently suffer from bronchitis and pneumonia Moreover, owing to the rapidity of the respirations and the congestion in the lungs, the condition is frequently misdiag

If there is severe pulmonary stenosis or functional pulmonary atresia, the infant will suffer from severe *attacks of paroxysmal dyspnea* as the ductus arteriosus undergoes obliteration When the pulmonary artery is of small caliber or the pulmonary orifice only slightly *stenotic*, although cyanosis is intense, the infant does better than would be expected from the intensity of the cyanosis

Growth and development are usually retarded

The appearance of the patient may suggest the existence of a cardiac abnormality The child who shows no cyanosis frequently has a *gracile habitus* because of the large pulmonary blood flow and the small and rapid systemic flow Consequently, even though the oxygen saturation of the arterial blood is nearly normal the systemic circulation is starved

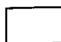
The presence or absence of cyanosis depends upon the volume of blood which reaches the lungs for oxygenation and the volume of venous blood which is directed into the aorta This, in turn, depends upon the size and position of the pulmonary artery and the relation of the tricuspid valve to the rudimentary outlet chamber When the pulmonary artery arises from the rudimentary outlet chamber, it is usually of small caliber and may be stenotic or even atretic at its base Under such circumstances only a small volume of blood reaches the lungs for oxygenation and a small volume of oxygenated blood is mixed with a large volume of venous blood Cyanosis is intense

If, however, there is a transposition of the great vessels, so that the pulmonary artery is given off the main ventricle, it is usually of normal caliber, and the aorta which arises from the rudimentary chamber, is of small caliber Under such circumstances a greater volume of blood goes to the lungs for oxygenation than to the systemic circulation Hence a large volume of oxygenated blood is mixed with a relatively small volume of venous blood There is usually no "visible" cyanosis

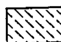
When there is a Lambert heart, that is, when a complete transposition of the great vessels is combined with an anomalous insertion of the tricuspid valve of such a nature that the major part of the venous blood from the right auricle is directed into the rudimentary outlet chamber and thence into the aorta, even though the pulmonary blood flow is large, the patient will show intense cyanosis

In brief, cyanosis is always present if there is decreased pulmonary blood flow but may be present when the pulmonary blood flow is *excessive* Under the latter circumstances the pulmonary artery arises posteriorly from the main ventricle and the occurrence of cyanosis is due to the anomalous insertion of the tricuspid valve which directs the blood from the right auricle into the rudimentary outlet chamber

DIAGRAM XV-4


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

Diagram XV-4

Single ventricle with a rudimentary outlet chamber from which the transposed aorta arises (Lambert heart)

In this malformation there are two auricles and a single ventricle with a rudimentary outlet chamber and the great vessels are transposed There may or may not be an auricular septal defect The mitral valve opens into the single ventricle and the chordae tendineae of the tricuspid valve are attached to the wall of the outlet chamber

Owing to the abnormality of the tricuspid valve most of the blood from the right auricle is directed into the rudimentary outlet chamber and thence is pumped out through the aorta to the systemic circulation and is again returned by the superior vena cava and the inferior vena cava to the right auricle The blood in the left auricle flows through the mitral valve into the common ventricle A large volume of the blood in the common ventricle is pumped out through the pulmonary artery to the lungs, where it is oxygenated and returned by the pulmonary veins to the left auricle thence it again flows into the common ventricle Nevertheless since there is a common ventricle some of the blood which flows through the tricuspid valve flows into the common ventricle and some of the blood in the common ventricle is pumped into the rudimentary chamber and thence out into the aorta Therefore although there is a transposition of the great vessels and a large proportion of the venous blood is pumped out into the aorta there is considerable mixing of venous and arterial blood in the common ventricle The origin of the pulmonary artery from the common ventricle causes severe pulmonary hypertension

Clinical diagnosis Cyanosis is intense and dates from birth Nevertheless there is some constant crossing of the two circulations in the common ventricle hence the patient is not as limited as might be expected from the intensity of the cyanosis Furthermore the condition is relatively stable and compatible with life for a number of years The child does not squat when tired The heart is slightly enlarged but there is no progressive cardiac enlargement There is absence of fullness of the pulmonary conus but the pulmonary vascularity is markedly increased and there may be a hilar dance The electrocardiogram frequently shows a discrepancy between the axis deviation in the standard leads and the evidence of ventricular hypertrophy in the unipolar precordial leads Pulmonary hypertension increases with age and usually causes progressive incapacity in early adult life

Clubbing of the extremities is directly proportional to the cyanosis If the patient shows no visible cyanosis and the red blood cell count is but slightly elevated, clubbing is absent If cyanosis and polycythemia are marked, clubbing develops at an early age

The exercise tolerance of the individual varies with the oxygen saturation of the arterial blood Nevertheless, since the patient is able to increase the pulmonary blood flow with exercise, his tolerance is better than is expected and he does not squat when tired For this reason, the possibility of a single ventricle should be considered when a child who is intensely cyanotic has a fair exercise tolerance Nevertheless, over a period of years the patient gradually becomes progressively more incapacitated

CARDIAC FINDINGS

Inasmuch as the structure of the heart is basically the same in all types of a single ventricle, the cardiac findings are similar

The heart at birth is normal in size The rate of enlargement is ordinarily so slow that only when the malformation is compatible with life for several years is there any demonstrable cardiac enlargement Furthermore, a balance is usually established, hence progressive cardiac enlargement, if it occurs at all, occurs extremely slowly

Murmurs are variable During the first months of life a murmur may or may not be present In childhood it is common to hear a precordial *systolic murmur* which may be maximal at the apex Since there is no shunting of blood from an area of high pressure to one of low pressure, the murmur lacks the rasping quality so characteristic of a small ventricular septal defect There may be both a systolic murmur and a low pitched, blurred mid diastolic murmur, thus the murmurs may resemble those of acquired rheumatic heart disease [3] Furthermore, a *gallop rhythm* is frequently heard The transmission of the systolic murmur is, however, not that of a mitral insufficiency and exercise fails to bring out a true crescendo presystolic murmur

The second sound over the pulmonary area is usually accentuated If the pulmonary artery is normally placed, the pulmonary hypertension causes accentuation of the second sound In cases of complete transposition of the great vessels, the aorta occupies the position of the normal pulmonary artery and hence the closure of the aortic valve causes the second sound to be louder to the left of the sternum than to the right Therefore, regardless of the presence or absence of cyanosis, the second sound at the base of the heart to the left of the sternum is usually accentuated It is also frequently reduplicated

Cardiac failure is prone to occur when there is excessive circulation to the lungs As the enlargement occurs extremely slowly, there may be chronic cardiac failure for many years In patients who show no cyanosis, the persistent cardiac failure may lead to the diagnosis of an insidious rheumatic infection Congestion of the lungs and engorgement of the liver commonly occur, and there may be edema and even ascites

The knowledge that the cardiac difficulty dates from infancy or early child hood, together with the absence of either history or symptoms of acute rheu matic fever, aids in the recognition of the nature of the cardiac difficulty Upon observation over a period of years there is a striking discrepancy between the apparently precarious compensation, combined with the poor quality of the heart sounds, and the absence of any progression of symptoms

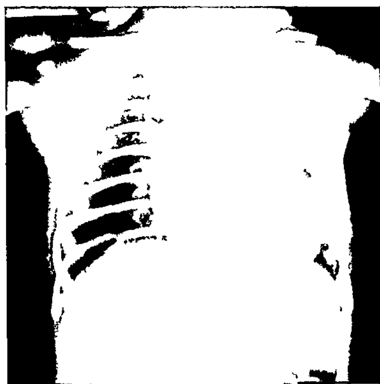
X RAY AND FLUOROSCOPIC FINDINGS

It is the shape of the heart which frequently gives the clue to the diagnosis The fact that the architecture of the heart is the same regardless of the position of the great vessels means that the contour of the heart is the same

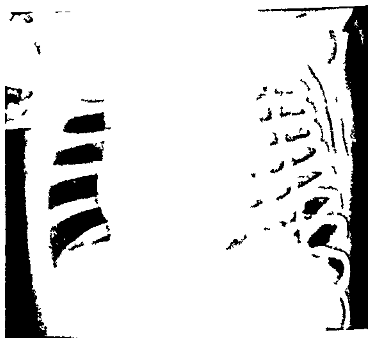
In this malformation there is a striking tendency for overexpansion of the lungs and depression of the diaphragm Consequently the chest is long and nar row and the contour of the heart is also long and narrow

Inasmuch as the rudimentary outlet chamber occupies the position of the normal outflow tract of the right ventricle, in early infancy there is a conspicuous bulge in this region Therefore, in the anterior posterior position, although the heart is not enlarged, there is prominence of the second curve to the left of the sternum, which suggests that the right ventricle is huge In reality there is no right ventricle, it is *only* a diminutive chamber It follows that the right ventricle cannot be enlarged The findings in the left anterior-oblique position confirm this fact the right ventricle does not project toward the anterior chest wall be yond the margin of the aorta These findings, as shown in Figures xv-4 and 5, demonstrate that the exaggeration of the shadow seen in the anterior posterior position in the region of the pulmonary conus is *not* caused by a huge right ven tricle it is in reality due to a rudimentary chamber

Although the contours of the heart are similar when both great vessels arise from the diminutive outlet chamber and when *only* one great vessel arises from the diminutive chamber and the other arises from the common ventricle (com pare Figures xv-4 6 and 7), the hilar shadows may differ greatly When the pulmonary artery arises from the common ventricle there is an excessive blood flow to the lungs, which in turn causes marked pulmonary congestion (see Fig

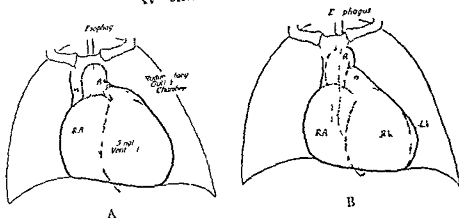


Anterior posterior position

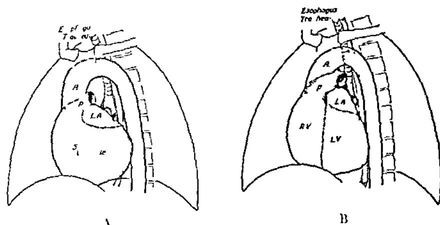


Left anterior-oblique position

FIGURE XX–4 Single ventricle with a rudimentary outlet chamber from which the pulmonary artery arises Infant



ANTERIOR POSTERIOR POSITION



LEFT ANTERIOR-OBLIQUE POSITION

FIGURE XV-7 (A) Single ventricle with a rudimentary outlet chamber and (B) normal heart Infant

ures XV-7, 8, and 11) *When the pulmonary artery is diminutive, the hilar shadows are minimal* Frequently, however, cardiac failure occurs early and by the time the patient comes under observation there may be congestion in the lungs

The contour of the heart changes appreciably with the growth of the individual As the child grows, the diaphragm descends still further and the apex of the heart rotates downward and inward When this occurs, the prominence of the pulmonary conus, which was caused by the rudimentary outlet chamber, disappears the contour of the heart comes to resemble that of a tetralogy of Fallot (compare Figures XV-8, 9 and 10 with Figures VI-17 and 18) or that of a transposition of the great vessels with pulmonary stenosis (see Figure X-26)

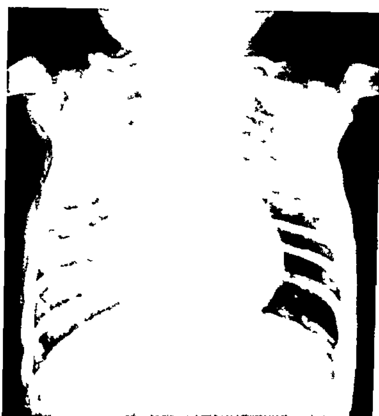


FIGURE XV–6 Single ventricle with a rudimentary outlet chamber from which both great vessels arise Infant

If the pulmonary artery arises from the common ventricle, the pulmonary artery is generally large and the pulmonary pressure is increased, consequently hilar pulsations are readily discernible It is the occurrence of greatly increased vascular shadows and a conspicuous hilar dance, combined with a concave curve at the base of the heart to the left of the sternum, which indicates that the pulmonary artery is transposed (see Figures XV–8 and 11) These two x rays show the basic contour of the heart in a child with a single ventricle and posteriorly placed pulmonary artery Nevertheless, the child whose x ray is shown in Figure XV–8 was intensely cyanotic, whereas the one whose x ray is shown in Figure XV–11 was not cyanotic

When the aorta arises from the common ventricle and the pulmonary artery is diminutive in size, there is no prominence of the pulmonary conus, and there is no increase in the pulmonary vascularity Indeed, the lungs may be exceptionally clear Under such circumstances the radiological findings, as shown in Figure XV–12, may closely resemble those of a transposition with pulmonary stenosis Compare Figures XV–12 and X–26

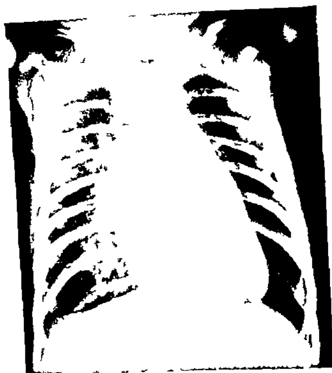


FIGURE XV-7 Single ventricle with a rudimentary outlet chamber from which the aorta arises (same patient as in Figure XV-1) Infant

Examination in the left anterior-oblique position may show that the straight anterior margin of the cardiac silhouette has disappeared by the time the patient reaches late childhood or early adolescence The vascular markings vary with the volume of the pulmonary blood flow, there is, however, always absence of the fullness of the pulmonary conus (see Figure XV-13 and also Figure XV-9) Examination in the right anterior-oblique position contributes little

Occasionally a transposition of the great vessels occurs in combination with pulmonary stenosis in which the aorta, as it arises anteriorly from the rudimentary outlet chamber, arches boldly to the left and causes the pulmonary conus to be extremely prominent and yet the lungs are remarkably clear (see Figure XV-14) This, too, causes the x ray contour of the heart to resemble that of a complete transposition of the great vessels combined with pulmonary stenosis (compare Figures XV-14 and X-27)

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is subject to wide variation The standard leads

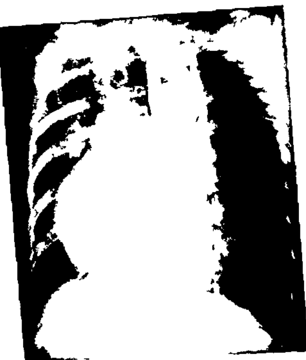


FIGURE xv–8 Single ventricle with a rudimentary outlet chamber from which the aorta arises (same patient as in Figure xv–2) Child

The pulmonary artery arises posteriorly from the main chamber

may show either a right axis deviation or a left axis deviation Although the course of the bundle of His must be grossly misplaced, it is a striking fact that there is usually no demonstrable disturbance of the intraventricular conduction time

Neill and Brink[4] have shown that, even in the presence of a left axis deviation, it is the exception, rather than the rule, for the unipolar precordial leads to show evidence of left ventricular hypertrophy in V_1, and furthermore a patient with a single ventricle may even have a so-called 'septal Q' wave in V_5 or V_6 Indeed, frequently when the standards show a slight left axis deviation, the unipolar precordial leads show evidence of right ventricular hypertrophy in V_6 which causes a deep S wave in all the precordial leads from V_1 to V_6 (see Figure xv–15) In other instances a right axis deviation is combined with left ventricular dominance in the unipolar precordial leads (see Figure xv–16) Thus, the electrocardiogram frequently shows a gross discrepancy between the findings in the standard leads and those in the precordial leads It is, however, possible to have a right axis deviation and right ventricular hypertrophy (see Figure xv–17)



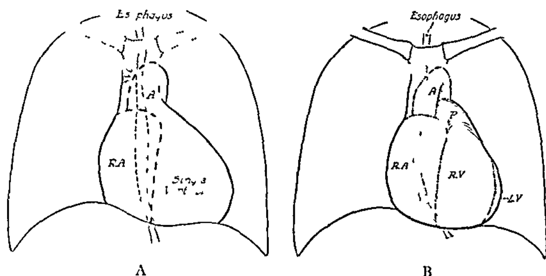
Left anterior-oblique position



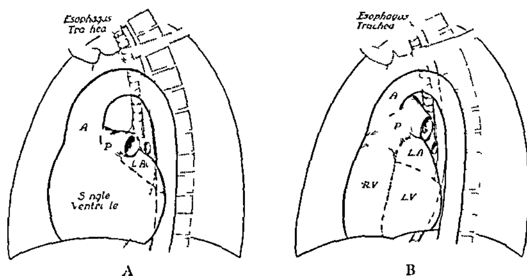
Right anterior-oblique position

FIGURE XV-9 Single ventricle with a rudimentary outlet chamber from which the aorta arises (same patient as in Figure XV-2) Child

The pulmonary artery arises posteriorly from the main chamber



ANTERIOR POSTERIOR POSITION



LEFT ANTERIOR-OBLIQUE POSITION

FIGURE XI-10 (A) Single ventricle with a rudimentary outlet chamber and (B) normal heart Child



Courtesy J Gl... dy Gl... dy a d W... d

FIGURE XI-II Single ventricle with a rudimentary outlet chamber from which the aorta arises Child

The pulmonary artery arises posteriorly from the main chamber

SPECIAL TESTS

The oxygen saturation of the arterial blood is never completely normal There is always admixture of the venous and arterial blood in the common ventricle When there is adequate circulation to the lungs and free admixture of the venous and arterial blood in the common ventricle, the oxygen saturation of the arterial blood *may be 94 per cent or higher* When the pulmonary blood flow is reduced, there is a proportionate reduction in the oxygen saturation of the arterial blood

Polycythemia is proportional to the oxygen unsaturation of the arterial blood Inasmuch as there is always some degree of oxygen unsaturation of the arterial blood, the red blood cell count, the amount of available hemoglobin, and the hematocrit reading are usually elevated The height of the red blood cell count varies with the oxygen unsaturation of the arterial blood

In the patient with free admixture of venous and arterial blood in the com



FIGURE XX-12 Single ventricle with pulmonary stenosis (same patient as in Figure XX-13) Adult

mon ventricle and adequate circulation to the lungs, although polycythemia may not be significant, the red blood cell count is usually a high normal Indeed, the absence of anemia presents a striking contrast to the frail build of the child

If the patient is cyanotic, polycythemia develops early Once the red blood cell count, the level of the available hemoglobin, and the hematocrit reading have risen to compensate for the reduction in the oxygen saturation of the arterial blood, polycythemia remains constant over a period of years The patient does not develop further hemoconcentration until the pulmonary hypertension becomes extreme

Cardiac catheterization may be of aid in diagnosis The most significant finding on cardiac catheterization is the marked increase in the oxygen content of the blood in the common ventricle as compared with that in the right auricle Bing considered that an increase of 5 volumes per cent in the oxygen content of the blood of the right ventricle, compared with that of the right auricle, is indicative of a common ventricle Such a finding is strong confirmatory evidence of a common ventricle, but such an increase can occur only if the difference between



Left anterior-oblique position

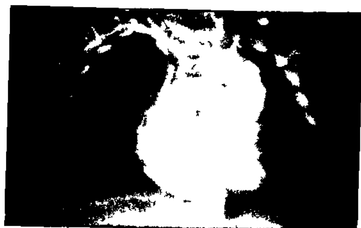


Right anterior-oblique position

FIGURE XV-13 Single ventricle with pulmonary stenosis (same patient as in Figure XV-12) Adult



Teleoroentgenogram



Angiocardiogram

FIGURE XV-14 Single ventricle with transposition of the great vessels, the aorta arising far to the left Infant

the oxygen content of the arterial blood and that of the venous blood is greater than 5 volumes per cent When there is tremendous pulmonary blood flow, the oxygen content of the venous blood may be so high that there is less than a 5 volume percentage difference between the arterial and venous blood Conversely, when there is severe pulmonary stenosis, the pulmonary blood flow may be so meager that the arterial oxygen saturation is too low to have a 5 volume percentage difference Furthermore, if a single ventricle occurs in combination with a single auricle, the increase in the oxygen content of the blood will occur in the auricle, and the oxygen content of the blood in the auricle and in the ventricle will be the same Therefore the finding of a marked increase in the oxygen content of the blood in the ventricle is suggestive evidence of a single ventricle, but its absence does not exclude the diagnosis

Angiocardiography shows simultaneous opacification of the aorta and the

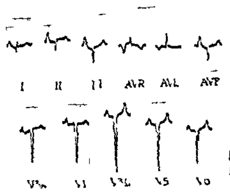


FIGURE XV-15 Single ventricle with pulmonary stenosis Adult

Note the deep S in all precordial leads left ventricular dominance in V_1 and right ventricular dominance in V_6

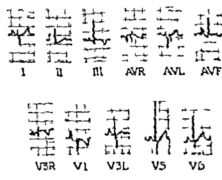


FIGURE XV-16 Single ventricle

Note the right axis deviation and the normal left ventricular dominance

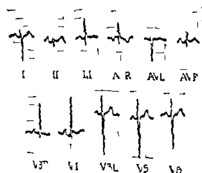


FIGURE XV-17 Single ventricle

Note the right axis deviation and right ventricular hypertrophy

pulmonary artery, it also clearly delineates the course of the aorta (see Figure XV–14) Furthermore, the dye promptly fills the entire ventricle, which usually has a smooth outline in contrast to the normal trabeculation of the right ventricle Such a contour does not exclude the possibility of a huge right ventricle, nor yet the absence of the right ventricle and the existence of a single ventricle, the left Nevertheless, when angiocardiography is undertaken to determine whether or not there is a single ventricle or two ventricles, the visualization of the right ventricle and later of the left ventricle demonstrates the existence of two ventricles

DIAGNOSIS

The diagnosis is based upon a combination of clinical, cardiological, and radiological findings, these vary with the age of the patient

In early infancy there is usually cyanosis of varying intensity, and a small heart with slight fullness of the pulmonary conus in the anterior posterior position, but no enlargement of the right ventricle in the left anterior oblique position Cyanosis may be so intense and the arterial oxygen saturation may be so low that the infant is in danger of dying from anoxemia, or cyanosis may be so slight that no cardiac abnormality is suspected and the condition is misdiagnosed as pneumonia

When there is pulmonary atresia, cyanosis is intense and the infant suffers from paroxysmal dyspnea, the heart is slightly enlarged and has a square contour similar to that of tricuspid atresia and the lungs are clear The discrepancy in the electrocardiogram between the standard leads and the unipolar precordial leads indicates that the condition is neither a tetralogy of Fallot nor a tricuspid atresia

In childhood there are at least three distinct clinical syndromes (1) when the pulmonary blood flow may be adequate or excessive with free admixture of venous and arterial blood in the common ventricle, (2) when the great vessels may be transposed and the pulmonary circulation is adequate but the occurrence of an abnormality of the tricuspid valve directs the venous blood into the aorta, (3) when the pulmonary artery may be small or the pulmonary valve stenotic and consequently pulmonary blood flow is reduced

In the first instance, *when the pulmonary blood flow is adequate or excessive and cyanosis is absent,* the diagnosis is based upon the finding of a patient of frail build with slight cardiac enlargement, with a precordial systolic murmur, and with a blurred mid diastolic murmur at the apex Fluoroscopy reveals in

creased hilar shadows The red blood cell count is normal but the oxygen saturation of the arterial blood is slightly reduced

In the second instance, *when cyanosis is intense and the pulmonary blood flow is adequate,* there is always polycythemia The heart is slightly enlarged and the hilar shadows are increased and may show a conspicuous hilar dance but the shadow at the base of the heart is concave The patient is less limited than might be expected from the intensity of the cyanosis When the pulmonary artery is huge, this malformation closely simulates that of a complete transposition of the great vessels with an enormous pulmonary artery (see Chapter X, Section B)

In the third instance, *when cyanosis is intense and the pulmonary blood flow is reduced* polycythemia develops early, the heart is only slightly enlarged but has a concave curve at its base to the left of the sternum, murmurs are variable, the pulmonic second sound is accentuated but the lungs are extremely clear, and the vascular markings are reduced

The infant does better than is anticipated from the intensity of the cyanosis The child can often play all day without undue fatigue In such instances the x ray may be similar to that of a tetralogy of Fallot but the child does not squat when tired The electrocardiogram usually offers a clue to the diagnosis, as there is frequently a striking discrepancy between the axis deviation in the standard leads and the evidence of left or right ventricular dominance in the unipolar precordial leads

DIFFERENTIAL DIAGNOSIS

In the absence of cyanosis a single ventricle requires differentiation from pneumonia and rheumatic heart disease In the presence of cyanosis, the condition requires differentiation from a tetralogy of Fallot from a tricuspid atresia, from a complete transposition of the great vessels with two ventricles and a posteriorly placed dilated pulmonary artery, and occasionally from a truncus arteriosus with a moderate reduction in the pulmonary blood flow

Pneumonia is a common error in diagnosis in infants When there is no murmur and the heart is of normal size, the occurrence of slight cyanosis and dyspnea combined with extensive pulmonary congestion, is strongly suggestive of pneumonia The failure to demonstrate pathogenic organisms in cultures from the nose and throat, combined with the contour of the heart in the x ray, offers a clue to the correct diagnosis Indeed, if the pathologist implies that the clinician has erroneously diagnosed pneumonia in a young infant who was

found to have a congenital malformation of the heart, the possibility of a bilocu late or triloculate heart should be immediately entertained

Rheumatic heart disease may be erroneously diagnosed in children who show no cyanosis Although the murmurs may superficially resemble those of a rheu matic myocarditis, careful analysis of murmurs will show that they are not char acteristic of that disease The systolic murmur is not well transmitted to the axilla The early diastolic murmur at the apex does not resemble a *presystolic murmur* The absence of a rheumatic history in a child with symptoms of long duration is also significant The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are usually normal or slightly elevated, whereas with a long standing rheumatic infection anemia is the rule Simple awareness of the possibility aids in the diagnosis of a single ventricle Fluoro-scopic examination corroborates the diagnosis Determination of the oxygen saturation of the arterial blood will reveal that it is not fully saturated Cardiac catheterization will show definite evidence of admixture of venous and arterial blood in the common ventricle

When the patient shows persistent cyanosis, x ray or fluoroscopy will show whether the pulmonary blood flow is increased or decreased When the pulmo-nary blood flow is increased, a single ventricle calls for differentiation from complete transposition of the great vessels with a dilated pulmonary artery, when decreased, a single ventricle requires differentiation from a tetralogy of Fallot, tricuspid atresia, a truncus arteriosus, and a complete transposition of the great vessels with pulmonary stenosis

Transposition of the great vessels combined with a greatly dilated pulmo nary artery and an intact ventricular septum gives identically the same x ray or fluoroscopic findings as does the Lambert heart The patient is usually more severely incapacitated if there are two ventricles because of the greater difficulty in the admixture of oxygenated and venous blood Cardiac catheterization is generally necessary in order to differentiate the two conditions Although the aorta may be readily entered, the catheter usually also slips into the common ventricle, in which the oxygen content of the blood will be far higher than it is in the right ventricle or in the aorta

Complete transposition of the great vessels with an auricular septal defect may occasionally show a tremendous increase in the oxygen content of the blood in the right ventricle and may be extremely difficult to differentiate from a Lambert heart Angiocardiography may be of aid by demonstration of an auricular defect, and also will usually demonstrate the existence of two ventricles and the late opacification of the pulmonary arteries

A tetralogy of Fallot may be easily confused with a single ventricle combined with pulmonary stenosis, primarily because of the contour of the heart A history of freedom from attacks of paroxysmal dyspnea and of absence of squatting is against the diagnosis of a tetralogy of Fallot Furthermore, on physical examination the systolic murmur usually is not as loud or as harsh as in a tetralogy of Fallot and the pulmonic second sound is accentuated The electrocardiogram also generally aids in the differentiation of the two conditions

A tricuspid atresia can usually be differentiated by the electrocardiogram, occasionally cardiac catheterization is necessary to determine whether or not a ventricle can be entered from the right auricle

A truncus arteriosus with moderately reduced pulmonary blood flow may be considered because of the absence of squatting and the relatively good exercise tolerance The continuous murmur audible over the lungs readily differentiates a truncus arteriosus from a single ventricle A truncus arteriosus may, however, be associated with a single ventricle, but if so, the single ventricle is mainly of academic interest, as all the blood from the common ventricle is pumped into the truncus arteriosus (see Chapter XIV)

Transposition of the great vessels combined with pulmonary stenosis is probably the most difficult of all conditions to differentiate from a single ventricle when the stenosed pulmonary artery arises from the common ventricle Regardless of whether the aorta arises normally from the mid portion of the right ventricle or whether it arches boldly to the left, the condition may simulate that of a single ventricle. Indeed, the demonstration of two ventricles by angiocardiography is virtually the only way to differentiate the two conditions with certainty

COMMONLY ASSOCIATED ANOMALIES

A single ventricle may occur in combination with tricuspid atresia Furthermore, this combination of anomalies may occur with or without a transposition of the great vessels, and with or without pulmonary stenosis Functionally the condition is closely similar to a tricuspid atresia and an underdeveloped right ventricle There are two auricles and a single ventricle The blood from the right auricle can leave only by way of the left auricle and thence the admixture of venous and arterial blood enters the common ventricle and is pumped out to both the systemic and pulmonary circulations

In the presence of a single ventricle, the blood usually circulates freely in the common chamber and less difficulty is encountered in the expulsion of blood from the rudimentary outlet chamber than through a small defect into a rudi

mentary right ventricle, hence the condition is more frequently compatible with life for a number of years than when tricuspid atresia is combined with a rudimentary right ventricle. Nevertheless, the two conditions merge into one another. It may be difficult even at autopsy to decide which condition is present.

The electrocardiogram provides the principal clinical clue to the differentiation of the two anomalies. Usually in a single ventricle the unipolar precordial leads do not show the evidence of left ventricular hypertrophy in V_1 which is the rule in tricuspid atresia. Neill and Brink[4] have, however, reported two cases in which the electrocardiograms were closely similar to that of tricuspid atresia.

A single ventricle may also occur in combination with mitral atresia as illustrated *in Figure* XI–*18*. When the mitral valve is atretic, a gross defect in the auricular septum is inevitable, as such an opening is the only way by which the blood can escape from the left auricle. The smaller the defect in the auricular septum, the greater is the enlargement of the left auricle. The occurrence of a single ventricle in combination with mitral atresia causes the pulmonary pressure to be excessively high because the pulmonary resistance is increased both by the high pressure in the left auricle (see Chapter XXX) and by the high pressure under which the blood is ejected to the lungs from the common ventricle. Consequently pulmonary hypertension is extreme and pulmonary congestion is severe.

TREATMENT

When a single ventricle occurs in combination with pulmonary stenosis, a systemic pulmonary anastomosis increases the circulation to the lungs and lessens cyanosis but may lead to progressive cardiac enlargement. A number of patients with the single ventricle and pulmonary stenosis die of cardiac failure within six to eight months after operation, others develop cardiac enlargement more slowly and may live for eight to ten years or longer. Nevertheless, some patients with a single ventricle are as greatly benefited by this operation as are those with a tetralogy of Fallot.

If the primary difficulty is due to excessive pulmonary blood flow, the production of moderate pulmonary stenosis may be of benefit to the patient. Obviously it is desirable that the pulmonary stenosis be sufficient to break the pulmonary pressure but not so great as to decrease pulmonary blood flow to the extent that it will produce cyanosis. Muller and Dammann[6] have developed a method for decreasing the size of the pulmonary artery by means of a "tucking" procedure. Such an operation may be of great value to the patient with a single ventricle.

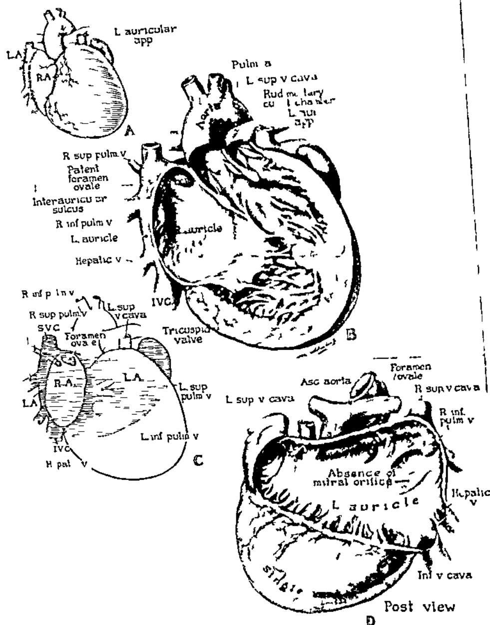


FIGURE XV-18 Single ventricle and mitral atresia

mentary right ventricle, hence the condition is more frequently compatible with life for a number of years than when tricuspid atresia is combined with a rudimentary right ventricle Nevertheless, the two conditions merge into one another It may be difficult even at autopsy to decide which condition is present

The electrocardiogram provides the principal clinical clue to the differentiation of the two anomalies Usually in a single ventricle the unipolar precordial leads do not show the evidence of left ventricular hypertrophy in V_1 which is the rule in tricuspid atresia Neill and Brink[4] have, however, reported two cases in which the electrocardiograms were closely similar to that of tricuspid atresia

A single ventricle may also occur in combination with mitral atresia as illustrated in Figure xi-18 When the mitral valve is atretic, a gross defect in the auricular septum is inevitable, as such an opening is the only way by which the blood can escape from the left auricle The smaller the defect in the auricular septum, the greater is the enlargement of the left auricle The occurrence of a single ventricle in combination with mitral atresia causes the pulmonary pressure to be excessively high because the pulmonary resistance is increased both by the high pressure in the left auricle (see Chapter xxx) and by the high pressure under which the blood is ejected to the lungs from the common ventricle Consequently pulmonary hypertension is extreme and pulmonary congestion is severe

TREATMENT

When a single ventricle occurs in combination with pulmonary stenosis, a systemic pulmonary anastomosis increases the circulation to the lungs and lessens cyanosis but may lead to progressive cardiac enlargement A number of patients with the single ventricle and pulmonary stenosis die of cardiac failure within six to eight months after operation, others develop cardiac enlargement more slowly and may live for eight to ten years or longer Nevertheless, some patients with a single ventricle are as greatly benefited by this operation as are those with a tetralogy of Fallot

If the primary difficulty is due to excessive pulmonary blood flow, the production of moderate pulmonary stenosis may be of benefit to the patient Obviously it is desirable that the pulmonary stenosis be sufficient to break the pulmonary pressure but not so great as to decrease pulmonary blood flow to the extent that it will produce cyanosis Muller and Dammann[6] have developed a method for decreasing the size of the pulmonary artery by means of a "tucking procedure Such an operation may be of great value to the patient with a single ventricle

from the rudimentary outlet chamber and the insertion of the chordae tendineae of the tricuspid valve are such that the blood from the right auricle is directed into the rudimentary outlet chamber, the resultant malformation is known as a Lambert heart

Inasmuch as there is but one ventricle, all the blood from both auricles flows into the common ventricle and the same admixture of venous and arterial blood is pumped out into the aorta and the pulmonary artery The oxygen saturation of the blood in the common ventricle depends upon the volume of venous blood which reaches the lungs for oxygenation If the total volume of blood directed to the lungs is small, the volume of oxygenated blood returned from the lungs is proportionally small and the oxygen content of the blood in the common ventricle is low Unless there is pulmonary stenosis, there is always pulmonary hypertension

The presence or absence of cyanosis depends upon the oxygen content of the blood in the common ventricle and the amount of venous blood directed into the aorta Usually there is free admixture of venous and arterial blood in the common ventricle and cyanosis is minimal or absent In the Lambert heart, although the oxygen content of the blood in the common ventricle is relatively high, the oxygen content of the blood in the rudimentary outlet chamber is low little oxygenated blood is directed into the aorta cyanosis is intense

A high red blood cell count is the rule In the absence of cyanosis the only evidence of polycythemia may be the persistence of the red blood cell count at a high normal level in a patient who looks chronically ill and suffers from repeated pulmonary infections When the patient shows persistent cyanosis, polycythemia is the rule

Clubbing is directly proportional to the polycythemia

The exercise tolerance of the individual varies with the oxygen saturation of the arterial blood nevertheless, in cyanotic patients it is better than is anticipated from the intensity of the cyanosis

The heart is usually but slightly enlarged The second sound is generally accentuated Murmurs are variable and may simulate those of a poorly functioning rheumatic heart

The x ray and fluoroscopic findings frequently give a clue to the diagnosis In infancy there is fullness of the pulmonary conus but no evidence of right ventricular enlargement in the left anterior-oblique position In older children, when the pulmonary artery is of small caliber and the lung fields are clear, the

If a child has a small pulmonary artery and suffers from reduced pulmonary blood flow under relatively high pulmonary pressure, it may be difficult, if not impossible, to help him by operation An anastomosis will only function provided the pulmonary pressure is lower than the systemic pressure Corrective surgery would necessitate the construction of a septum within the ventricles in order to reduce the pressure with which blood is directed to the lungs In addition, if there is but a single atrioventricular valve, it will be necessary to reconstruct the atrioventricular orifice so that there is both a mitral and a tricuspid valve

If the patient has a Lambert heart, the creation of an auricular septal defect may increase the amount of oxygenated blood directed to the aorta but it will not alleviate the pulmonary hypertension

PROGNOSIS

The prognosis is guarded *In the absence of cyanosis and excessive pulmonary blood flow,* the patient is very susceptible to pneumonia, which is a constant menace to life *In the presence of cyanosis and excessive pulmonary blood flow,* the increasing pulmonary hypertension increases the volume of blood directed to the systemic circulation and consequently the condition of the patient remains relatively stationary for ten or twenty years Nevertheless, in both instances the pulmonary hypertension will eventually render the condition incompatible with life

In the presence of pulmonary stenosis there is always cyanosis Any operation which increases the circulation to the lungs may help Although some patients are greatly helped by operation, it must be admitted that the human circulation and metabolism are based on a four chambered heart and the separation of the two circulations A single ventricle is not an efficient mechanism and consequently the prognosis is guarded

SUMMARY

A single ventricle with a rudimentary outlet chamber represents an extremely early arrest in the development of the heart Instead of two ventricles, there is but a single ventricle into which both the mitral and tricuspid valves open In the position normally occupied by the outflow tract of the right ventricle, there is a rudimentary chamber from which one or both the great vessels arise The great vessels may or may not be transposed When the aorta arises

References

1 Taussig H B Cardiovascular anomalies a single ventricle with a diminutive outlet chamber J Tech Methods 19 120–128 1939

2 Lambert E C Single ventricle with a rudimentary outlet chamber case report Bull Johns Hopkins Hosp 88 231–238 1951

3 Glendy M M R E Glendy and P D White Cor biatriatum triloculare, case report Am Heart J 28 395–401 1944

4 Neill C A and A J Brink Left axis deviation in tricuspid atresia and single ventricle the electrocardiogram in 36 autopsied cases Circulation 12 612–619 1955

5 Bing R Personal communication

6 Muller W H Jr and J F Dammann, Jr The treatment of certain congenital malformations of the heart by the creation of pulmonic stenosis to reduce pulmonary hypertension and excessive pulmonary blood flow Surg Gynec & Obst 95 213–219 1952

contour of the heart may closely simulate that of a tetralogy of Fallot When the pulmonary artery *is of normal size and posteriorly placed,* the hilar vascularity is increased and there is a conspicuous hilar dance

The electrocardiogram is subject to great variation There may be a right or a left axis deviation, but usually there is a discrepancy between the axis deviation in the standard leads and the evidence of hypertrophy in the unipolar precordial leads

Cardiac catheterization characteristically shows a marked increase in the oxygen content of the blood in the common ventricle in comparison with that in the right auricle The systolic pressure in the common ventricle is always the same as the systolic pressure in the systemic circulation, consequently, unless there is pulmonary stenosis, there is always pulmonary hypertension

Angiocardiography shows simultaneous opacification of the aorta and the pulmonary artery but is not of great aid in the diagnosis of the structure of the ventricle

The diagnosis is based upon the clinical findings and the contour of the heart in the x ray combined with electrocardiographic findings of a discrepancy between the axis deviation in the standard leads and the ventricular dominance in the precordial leads The diagnosis may be substantiated by cardiac catheterization

In the absence of cyanosis, the condition must be differentiated from pneumonia in infancy and from rheumatic heart disease in childhood If there is *persistent cyanosis, the malformation may resemble a tetralogy* of Fallot The Lambert heart requires differentiation from a complete transposition of the great vessels with a dilated pulmonary artery and occasionally from a truncus arteriosus

Treatment is not satisfactory If cyanosis is absent and the pulmonary blood flow excessive, a tuck in the pulmonary artery may be of great help If cyanosis is intense and the pulmonary blood flow excessive, the creation of an auricular defect may help When the pressure is the same in both great vessels, an anastomosis will not function, hence it may be impossible to help a child with persistent cyanosis and a relatively small pulmonary artery

The prognosis is guarded Many infants with intense cyanosis and minimal pulmonary blood flow *die within the first few months Patients with a moderate* reduction in pulmonary blood flow may live to childhood Most patients with adequate pulmonary blood flow, with or without cyanosis, survive to adult life

as to cause high output cardiac failure in the neonatal period, are comparatively rare The sequence of events is illustrated by Case XVI-1 at the end of the chapter

Cerebral arteriovenous aneurysms unfortunately, are not rare Usually they are small and asymptomatic until early adult life The outstanding physical finding is a localized bruit Such aneurysms may bleed before they finally rupture or they may remain entirely unsuspected until the young adult suddenly develops a severe headache and dies of a massive cerebral hemorrhage

The frequency of congenital cerebral arteriovenous aneurysms in patients with malformations of the heart is difficult to ascertain Systolic bruits over the skull are extremely common in patients with cardiac malformations Certainly some of these bruits are the result of the bone transmission of cardiac murmurs Continuous murmurs over the skull are not infrequently heard in patients with persistent cyanosis Many such patients are entirely asymptomatic Inasmuch as extensive investigation of cerebral abnormalities is not without risk, it has not been our policy to undertake such studies in an asymptomatic person unless there were some localizing signs Nevertheless, in a number of instances a continuous murmur over the skull may be indicative of a cerebral arteriovenous aneurysm[4]

Coronary arteriovenous aneurysms are fundamentally similar to other congenital arteriovenous aneurysms but as they represent an anomaly of the coronary arteries they are discussed in Chapter XXIX

B *Pulmonary Arteriovenous Aneurysms*

Arteriovenous aneurysms in the lungs have been reported under various names hemangiomas of the lung, cavernous angiomas, and pulmonary arteriovenous aneurysms The first unequivocal case is credited to Rodes,[5] in 1938 The condition was reviewed and reported as a new surgical disease by Watson[6] in 1947 Two years later the radiological findings were reported by Grishman et al [7] For a detailed review the reader is referred to the reports by Sloan and Cooley [8, 9]

Almost all arteriovenous aneurysms are congenital in nature Some aneurysms undoubtedly increase in size as the patient grows and therefore become clinically manifest in late childhood or early adult life In former years the condition was frequently misdiagnosed as a congenital malformation of the heart and considered inoperable With the advent of cardiac surgery, many of these patients, even though asymptomatic, have sought medical attention and have been helped

NATURE OF THE MALFORMATION

The malformation consists of an abnormality in the pulmonary vascular bed

CHAPTER XVI

ARTERIOVENOUS ANEURYSMS

AN arteriovenous aneurysm is defined as a blood containing tumor or cavity with both a systemic and a venous connection. Although some are the result of trauma, this chapter is concerned with those which are congenital in origin. It seems probable that many, if not all hemorrhagic telangiectases, hemangiomas, and cavernous angiomas are fundamentally arteriovenous aneurysms. Indeed, the frequent association of hemorrhagic telangiectases and pulmonary arteriovenous aneurysms suggests that both are essentially of the same nature.[1]

Arteriovenous aneurysms may occur anywhere in the systemic or the pulmonary circulation. They may occur in the extremities, in the brain, or even in the myocardium, and they also occur in the lungs. When it is recalled that arteries and veins develop independently, it is not surprising that anomalous connections between the arteries and veins may occur anywhere in the body, and, furthermore, if the development is such that an arteriovenous aneurysm occurs in one place, others may occur in other locations. Therefore, although there may be only a single arteriovenous aneurysm, frequently they are multiple. The size of the anomalous connection varies greatly; it may be large or small.

A. *Systemic Arteriovenous Aneurysms*

Systemic arteriovenous aneurysms vary in size. Some are minute and entirely asymptomatic. Others may be large. When an arteriovenous aneurysm occurs in the systemic circulation, blood is shunted under systemic pressure into a vein; consequently the volume of the shunt is usually large. The blood so shunted is fully oxygenated. *It follows that a large volume of fully oxygenated blood is returned to the right auricle,* thence it flows to the right ventricle and is recirculated through the lungs. The work of the right side of the heart is increased. A large arteriovenous fistula may greatly increase the work of the heart and may lead to cardiac failure due to the high output of the heart.

The hereditary nature of hemorrhagic telangiectases was described by Osler in 1901 and by Weber[3] in 1907. This syndrome is now known as Rendu-Osler-Weber's disease. Such telangiectases may occur in almost any organ of the body. The problem is primarily medical and hence beyond the scope of this book.

culation The shunt, however, occurs in the lungs Consequently it occurs in an area of low pressure There is no tendency for the patient to develop pulmonary hypertension The only effect on the circulation is that the arterial blood is not fully oxygenated and the work of the heart is increased

CLINICAL FINDINGS

The patient frequently remains asymptomatic for many years If he has a complaint it is usually that his color is not normal

Cyanosis ordinarily dates from birth The intensity of the cyanosis depends upon the size and number of the arteriovenous aneurysms in the lungs Occasionally, although some degree of oxygen unsaturation of the arterial blood has been present from birth the volume of reduced hemoglobin in the circulating blood is insufficient to cause visible cyanosis This is notably true in infancy, because anemia is common

Polycythemia develops secondary to the oxygen unsaturation of the arterial blood With the development of polycythemia, cyanosis becomes more readily apparent

Clubbing of the extremities may eventually appear

Dyspnea is not striking but there may be exertional dyspnea Although some blood is shunted through the lungs without being oxygenated, a large portion of the pulmonary circulation is normal hence the patient is able to increase his pulmonary blood flow with exercise Consequently dyspnea is minimal The patient s exercise tolerance is better than might be expected from the degree of cyanosis

Anemia may be the presenting complaint Many patients with pulmonary arteriovenous aneurysms also have systemic arteriovenous aneurysms or hemorrhagic telangiectases If these are located in the gastrointestinal tract or in other mucous membranes, persistent oozing and bleeding may lead to severe anemia

Hemorrhagic telangiectases and hemangiomas are extremely common in patients with pulmonary arteriovenous aneurysms Therefore a search for these should always be made The converse is also true The presence of a systemic arteriovenous aneurysm should suggest the possibility that the cyanosis may be due to an aneurysm in the pulmonary circulation

Epistaxes are common They are in all probability due to minute arteriovenous aneurysms in the nose

Hemoptyses are a serious and late complication and may vary from repeated small hemorrhages to a massive fatal hemorrhage

of such a nature that there is free and broad communication between some of the arteries and veins within the lungs There may be single or multiple aneurysms, they may be large or small, such aneurysms may be localized in one lobe or scattered throughout both lungs The aneurysm usually involves the smaller branches of the pulmonary artery, which, instead of subdividing and branching into the capillary bed, open into a sacular cavity and this in turn communicates directly with the veins Frequently there are numerous cavities of various sizes, occasionally the lung is riddled with small cavities and often both lungs are involved In rare instances an aneurysm may occur between the bronchial artery and the pulmonary vein or may even occur in an anomalous artery leading to the lungs, as in the case reported by W L Watson,[6] in which there was an arteriovenous aneurysm between an aberrant artery from the thoracic aorta and what appeared to be a pulmonary vein The author has seen one patient in whom the anomalous branch from the pulmonary artery opened directly into the left auricle (see Case XVI-2 at end of chapter)

Pulmonary arteriovenous aneurysms tend to increase in size just as other aneurysms do The increase in size, however, occurs more gradually in a low pressure area than in a high pressure area For this reason it may require years for the increase in the size of aneurysms to become apparent

COURSE OF THE CIRCULATION

The blood from the right auricle flows into the right ventricle and is pumped out in the normal fashion to the lungs Part of the blood circulates normally through the normal lung, where it is oxygenated, and returned in the normal fashion to the left auricle A varying volume of blood passes directly from the pulmonary artery through the arteriovenous aneurysm to the pulmonary veins Consequently a mixture of oxygenated and unoxygenated blood is returned by the pulmonary veins to the left auricle This admixture of venous and arterial blood flows into the left ventricle, is pumped out through the aorta to the systemic circulation, and is returned to the right auricle in the normal manner There the cycle starts again (see Diagram XVI-1)

PHYSIOLOGY OF THE MALFORMATION

The shunting of the venous blood from the pulmonary artery into a pulmonary vein without passing through the capillaries prevents the oxygenation of the blood in that area of the lung Venous blood is returned to the left auricle and thence it flows to the left ventricle and is pumped out into the systemic cir

culation The shunt, however, occurs in the lungs Consequently it occurs in an area of low pressure There is no tendency for the patient to develop pulmonary hypertension The only effect on the circulation is that the arterial blood is not fully oxygenated and the work of the heart is increased

CLINICAL FINDINGS

The patient frequently remains asymptomatic for many years If he has a complaint, it is usually that his color is not normal

Cyanosis ordinarily dates from birth The intensity of the cyanosis depends upon the size and number of the arteriovenous aneurysms in the lungs Occasionally, although some degree of oxygen unsaturation of the arterial blood has been present from birth, the volume of reduced hemoglobin in the circulating blood is insufficient to cause visible cyanosis This is notably true in infancy, because anemia is common

Polycythemia develops secondary to the oxygen unsaturation of the arterial blood With the development of polycythemia, cyanosis becomes more readily apparent

Clubbing of the extremities may eventually appear

Dyspnea is not striking but there may be exertional dyspnea Although some blood is shunted through the lungs without being oxygenated, a large portion of the pulmonary circulation is normal, hence the patient is able to increase his pulmonary blood flow with exercise Consequently dyspnea is minimal The patient's exercise tolerance is better than might be expected from the degree of cyanosis

Anemia may be the presenting complaint Many patients with pulmonary arteriovenous aneurysms also have systemic arteriovenous aneurysms or hemorrhagic telangiectases If these are located in the gastrointestinal tract or in other mucous membranes persistent oozing and bleeding may lead to severe anemia

Hemorrhagic telangiectases and hemangiomas are extremely common in patients with pulmonary arteriovenous aneurysms Therefore a search for these should always be made The converse is also true The presence of a systemic arteriovenous aneurysm should suggest the possibility that the cyanosis may be due to an aneurysm in the pulmonary circulation

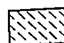
Epistaxes are common They are in all probability due to minute arteriovenous aneurysms in the nose

Hemoptyses are a serious and late complication and may vary from repeated small hemorrhages to a massive fatal hemorrhage

DIAGRAM XVI-1


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

DIAGRAM XVI–1

Multiple arteriovenous aneurysms

This *malformation* concerns the pulmonary arteries and pulmonary veins, the heart is itself normally formed The major part of the pulmonary vascular bed is normal but scattered throughout the lung there are arteriovenous aneurysms connecting some of the smaller branches of the pulmonary arteries with the pulmonary veins

The blood from the right auricle flows into the right ventricle and thence is pumped out into the pulmonary artery to the lungs Part of the blood circulates through the normal portions of the lungs where it is oxygenated, and is returned in the normal manner by the pulmonary veins to the left auricle. Part of the blood which is pumped to the lungs flows through an arteriovenous aneurysm The blood which circulates through such an aneurysm never reaches the pulmonary capillaries and hence is returned to the left auricle unoxygenated Thus a mixture of oxygenated and venous blood flows through the pulmonary veins to the left auricle thence the blood flows into the left ventricle and is pumped out through the aorta to the body and returned by the superior vena cava and the inferior vena cava in the normal manner to the right auricle There the cycle starts again

Clinical diagnosis Cyanosis is persistent and increases as the arteriovenous aneurysm increases in size. The patient is usually asymptomatic The heart is normal in size No murmurs are audible over the precordium but a continuous murmur is generally audible over the aneurysm If the aneurysms are large or increase in size, the patient is in danger of severe, even fatal pulmonary hemorrhages

Cerebral disturbances may occur

Transitory neurological disturbances are relatively frequent These may be due to anoxemia, polycythemia, minute thromboses, or to bleeding from minute cerebral aneurysms

Cerebral thromboses may occur in conjunction with small cerebral arteriovenous aneurysms

CARDIAC FINDINGS

The heart is generally normal in size Usually no murmurs are audible over the precordium The *pulmonic second sound* may be slightly accentuated A *soft continuous murmur* is usually audible over the area of the lung where there is a relatively large arteriovenous aneurysm

X RAY AND FLUOROSCOPIC FINDINGS

The x ray generally gives the clue to the diagnosis [8, 9] Although the heart is essentially normal in size and shape, an abnormal shadow is usually visible somewhere in the lungs (see Figure xvi–1) These abnormal shadows can frequently be seen upon fluoroscopy, indeed, intrinsic pulsations of the shadows may even be discernible When an arteriovenous aneurysm is suspected but not clearly evident, the patient should be carefully examined in all positions, because the aneurysm may be hidden behind the cardiac shadow

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram usually shows a balanced axis and no evidence of right ventricular hypertrophy

SPECIAL TESTS

The oxygen saturation of the arterial blood is reduced Nevertheless, the degree of unsaturation is less than is anticipated from the intensity of the cyanosis Furthermore, the arterial oxygen saturation falls but slightly, if at all, with exercise

Cardiac catheterization studies are frequently misleading The Fick principle is no longer valid because the blood returned to the left auricle is not fully saturated The pressures are normal, the resistance in the pulmonary vascular bed is normal [10]

Angiocardiography is of great diagnostic aid, it not only opacifies the princi



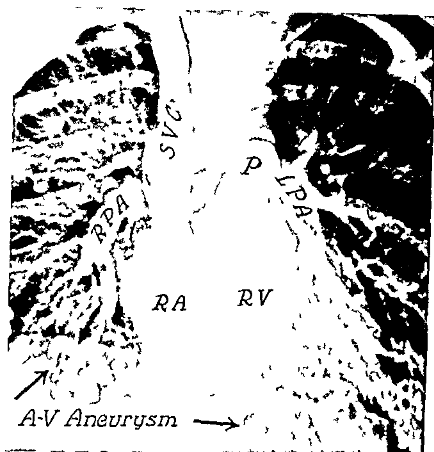
FIGURE XVI-1 Multiple pulmonary arteriovenous aneurysms (*same patient as in Figure XVI-2*) Adult

pal arteriovenous aneurysm but may also demonstrate additional unsuspected aneurysms (see Figure XVI-2) Angiocardiography is, however, not without danger, as the dye is pooled in the aneurysms Deaths have been reported from this procedure [11]

Laminograms are of great aid Such films give an indication of the size and position of the aneurysm and may reveal the tortuous vessels in and around it Planograms do not cover the entire lung and consequently other aneurysms may be missed Nevertheless, planography is a simple and safe procedure which aids in localization of the mass

DIAGNOSIS

The diagnosis should be suspected in a patient with persistent cyanosis and no limitation of activity and no complaints Physical examination reveals no cardiac abnormality A localized continuous murmur over the lungs is strong presumptive evidence in favor of the diagnosis The diagnosis is substantiated by



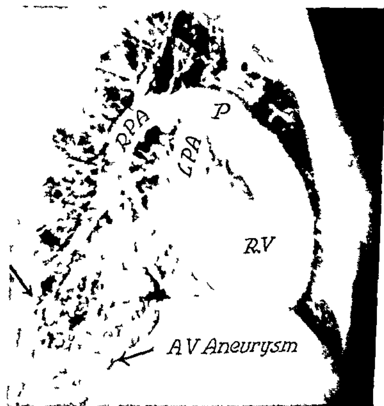


FIGURE XVI-2 Multiple pulmonary arteriovenous aneurysms (same patient as in Figure XVI-1) Adult

x ray or fluoroscopy and may be confirmed by an angiocardiogram or by a laminogram

DIFFERENTIAL DIAGNOSIS

The condition requires differentiation from congenital methemoglobinemia, from anomalous communication between the pulmonary artery and the left auricle, and also from congenital malformations of the heart, notably a truncus arteriosus, or localized stenoses in the branches of the pulmonary artery

Congenital methemoglobinemia is to be suspected in a patient with an absolutely normal heart, normal lungs, and persistent cyanosis which dates from birth The usual story is that the abnormal color was noted at birth Nevertheless the infant gains and thrives so well that the cyanosis causes little or no concern Even if a physician is consulted, the infant is doing so well that further investigations are seldom advised It is the normality of growth and development in the presence of persistent cyanosis that gives the clue to the diagnosis Occasionally the condition is familial and the knowledge that other members of the family have also shown an abnormal color aids in the diagnosis

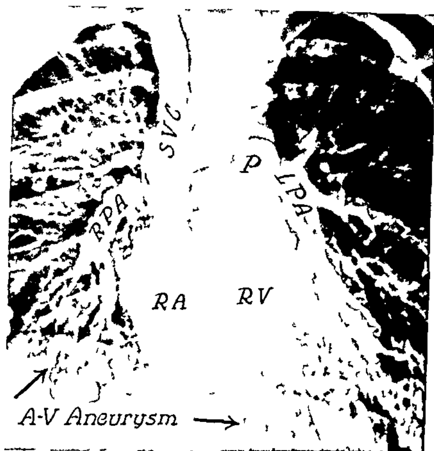
Methemoglobin poisoning may also be caused by the ingestion of nitrites Such poisoning is occasionally due to the use of contaminated well water in a baby s formula[1, 13]

The differentiation of cyanosis caused by methemoglobin from that caused by the presence of reduced hemoglobin in the circulating blood can easily be determined by the following test A drop of the patient s blood is placed on a piece of paper beside a drop of one s own blood When the two samples are stirred, the normal venous blood becomes bright red whereas the blood which contains methemoglobin is unable to combine with the oxygen in the air and therefore retains its dark color[14]

The diagnosis is definitively established by spectroscopic analysis of the blood and by the demonstration of the band characteristic of methemoglobin Cyanosis can be relieved by the injection of methylene blue and the patient can be cured by the continuous use of massive doses of ascorbic acid

Abnormal vascular communication between the pulmonary artery and the left auricle may simulate the picture of a pulmonary arteriovenous aneurysm (see Case XVI-2 at end of chapter)

A truncus arteriosus also causes persistent cyanosis and a continuous murmur over the lung Examination of the heart usually reveals a harsh systolic murmur and accentuation of the second sound over the base of the heart The contour of



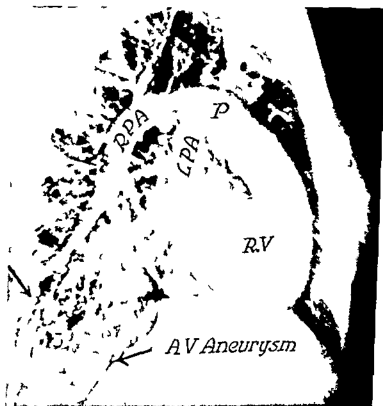


FIGURE XVI-2 Multiple pulmonary arteriovenous aneurysms (same patient as in Figure XVI-1) Adult

Pulmonary arteriovenous aneurysms are *frequently* multiple Inasmuch as venous blood from the pulmonary artery is shunted into the pulmonary vein without passing through the capillaries of the lungs the blood returned to the left auricle is not fully saturated Consequently the arterial blood is not fully saturated

The patient is cyanotic but usually asymptomatic The heart is normal in size The outstanding finding is a localized continuous murmur audible over the lung at the site of the aneurysm The aneurysm is frequently visible upon x ray or fluoroscopy and can almost invariably be delineated by angiocardiography or laminography

The condition calls for differentiation from congenital methemoglobinemia and other conditions which cause a continuous murmur over the lungs, notably peripheral pulmonary stenoses and a truncus arteriosus

The condition can be corrected by surgery

Prognosis is guarded but can be greatly improved by surgery

Unusual Illustrative Cases

CASE XVI-1 B S (Harriet Lane Home No A-98289) White male. Admitted at thirty six hours of age because of a mass in the right temporal region and severe cardiac failure

History The baby was born at the Lutheran Hospital He cried immediately and breathed well and then developed apnea He was given positive pressure oxygen and respiration was restored Shortly after birth the nurse noted a mass in the right temporal region Examination of the mass revealed a thrill and a bruit This mass rapidly increased in size Respiration became rapid Because of the dyspnea an x ray of the heart was taken at eighteen hours of age this revealed cardiac enlargement The baby was given digitalis

Physical examination The infant was dyspneic and cyanotic The respiratory rate was 200 per minute The heart was greatly enlarged and there was a gallop rhythm The liver extended to the umbilicus There were rales in the lungs

Over the right temporal region there was a large, soft pulsating mass approximately 5 cm in diameter In the center of this area there was no bony structure consequently the mass lay beneath the skin Around the periphery of the mass a delicate rim of the bone could be palpated A thrill was palpable over the mass and on auscultation there was a continuous bruit with a systolic accentuation Both the murmur and the thrill could be eliminated by pressure over the carotid artery

Diagnosis The infant obviously had a large arteriovenous aneurysm involving the carotid artery and high-output cardiac failure

the heart and the electrocardiogram are also abnormal Cardiac catheterization shows that the aorta arises in part or entirely from the right ventricle (see Chapter XIV)

Localized constrictions in one or more of the branches of the pulmonary artery are also a possible cause of a continuous murmur over the lungs The condition by itself does not produce cyanosis Nevertheless, peripheral pulmonary stenoses frequently occur in combination with some malformation of the heart which does cause cyanosis Therefore if a continuous murmur is due to peripheral stenoses and there is cyanosis, examination of the heart should reveal some abnormality

TREATMENT

Excision of the affected area offers the only possible cure Lobectomy or pneumonectomy is indicated if the major pathology lies in one lung, as there is real danger of serious, even fatal, hemorrhage

PROGNOSIS

Pulmonary arteriovenous aneurysms may be compatible with life for many years If the aneurysms are large and localized, or if the patient has had hemoptyses, operation is clearly indicated Provided the mortality rate from lobectomy or pneumonectomy is low, operation is indicated for any patient with a pulmonary arteriovenous aneurysm sufficiently large to cause cyanosis, as there is always the potential danger of rupture of the aneurysm

SUMMARY

An arteriovenous aneurysm is a blood containing tumor or cavity with both an arterial and a venous connection It seems probable that hemorrhagic telangiectases, hemangiomas, and cavernous angiomas are all basically arteriovenous aneurysms Such arteriovenous aneurysms may occur in either the systemic or the pulmonary circulation Some are single, many are multiple

Systemic arteriovenous aneurysms may be caused by trauma A large congenital arteriovenous aneurysm may cause high output cardiac failure in the neonatal period Small aneurysms are compatible with life for many years Such aneurysms in the brain are not rare and occasionally an aneurysm occurs in the myocardium between the coronary artery and the coronary vein The outstanding physical finding is a continuous murmur which is located over the area of the aneurysm

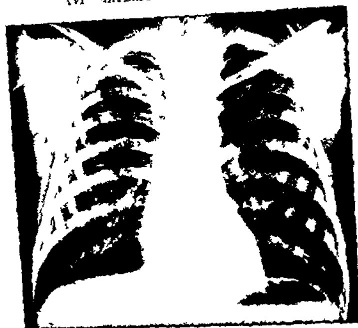


FIGURE XVI-3 Anomalous communication between the right pulmonary artery and the left auricle (Case XVI-2) Child

Treatment The patient was operated on by Dr Alfred Blalock on July 14 1948 The chest was entered on the right. A large soft, pulsating mass was found in the posterior part of the right lung The right pulmonary artery was exposed and a large thin walled vessel was found which arose from the posterior aspect of the right pulmonary artery and extended to the mass Further dissection showed that an anomalous vessel extended from the mass to the left auricle The anomalous vessel was ligated and divided The patient s color immediately improved No further surgery was attempted

Postoperative course The patient made an excellent recovery The pulsating mass in his forehead shrank until it became almost invisible One year and again three years after the operation the patient was re-examined Red blood cell count 4 6 million hemoglobin 15 gm hematocrit reading 39 oxygen saturation 93 per cent He remained well

Comment This patient had an anomalous vessel which arose from the right pulmonary artery and extended to a vascular mass which communicated directly with the left auricle This probably represented a developmental anomaly of the pulmonary vascular bed Functionally it was similar to an arteriovenous aneurysm Furthermore the condition was associated with a systemic arteriovenous aneurysm on his forehead

Treatment Cedilanid was given in the dispensary with but temporary improvement The only possible hope for the infant's life was ligation of the right external jugular artery and the carotid artery This operation was rapidly performed by Dr David Sabiston The bruit and thrill were entirely eliminated The heart rate immediately slowed to 60 beats per minute and continued to slow for the next half hour Thereafter the heart action ceased

Autopsy A partial autopsy was performed at the Lutheran Hospital A large vessel was found which connected the right external carotid artery, the middle meningeal artery, and the right lateral dural sinus

Case xvi–2 S X (Harriet Lane Home, No A–63444, Johns Hopkins Hospital, No 467587) White male First seen at the Harriet Lane Home in 1948 at fifteen years of age * The chief complaint was abnormality of color

History Cyanosis was first noted at five years of age, when the patient was seen at another hospital At this examination he was found to have a pulsating lump on his forehead During the ensuing ten years the mass slowly increased in size and cyanosis deepened The patient s activity, however, was but slightly restricted Running caused dyspnea but he could walk a mile and had taken short Scout hikes

Physical examination Temperature 37 1°C , pulse 92 per minute respirations 20 per minute, blood pressure 120/80 mm of mercury

The patient was a small undernourished lad, intensely cyanotic with marked clubbing of the extremities and a small pulsating mass on his forehead The heart was normal in size The heart sounds were of good quality, no murmur was heard The remainder of the physical examination was negative

Laboratory data Red blood cell count 9 5 million hemoglobin 25 gm hematocrit reading 77

Electrocardiogram Normal sinus mechanism There was a slight right axis deviation but the unipolar precordial leads were normal

Teleoroentgenogram The contour of the heart was normal the vascular markings, however, were increased and showed minimal pulsations in the hilar shadows (see Figure xvi–3)

Angiocardiography Immediately after the dye entered the pulmonary artery a relatively large, rounded area was opacified in the right hilum and a fraction of a second later the dye was seen in the left auricle This finding suggested the presence of a large arteriovenous aneurysm in the right lung which poured blood directly into the left auricle (see Figure xvi–4)

Diagnosis Arteriovenous aneurysm in the right lung

* This case was also reported by Sloan and Cooley [8]

References

1 Brink A J Telangiectasis of lungs, with 2 case reports of hereditary haemorrhagic telangiectasia with cyanosis Quart. J Med 19 239-248 1950
2 Osler W On a family form of recurring epistaxis associated with multiple telangiectases of the skin and mucous membranes Bull Johns Hopkins Hosp 12 333-337, 1901
3 Weber F P Multiple hereditary developmental angiomata (telangiectases) of the skin and mucous membranes associated with recurring hemorrhages Lancet 2 160-162 1907
4 Berthrong M and D C Sabiston, Jr Cerebral lesions in congenital heart disease a review of autopsies in 162 cases Bull. Johns Hopkins Hosp 89 384-406 1951
5 Rodes C B Cavernous hemangiomas of the lung with secondary polycythemia J.A.M.A 110 1914-1915 1938
6 Watson W L Pulmonary arteriovenous aneurysm—a new surgical disease Surgery 22 919-929 1947
7 Grishman A M H Poppel R. S Simpson, and M L Sussman Roentgenographic and angiocardiographic aspects of (1) aberrant insertion of pulmonary veins associated with interatrial septal defect and (2) congenital arteriovenous aneurysm of the lung Am J Roentgenol 62 500-508 1949
8 Sloan R D and R N Cooley Congenital pulmonary arteriovenous aneurysm Am J Roentgenol 70 183-210 1953
9 Cooley R N and R D Sloan Radiology of the Heart and Great Vessels Baltimore Williams & Wilkins 1956
10 Friedlich A., R. J Bing and S G Blount Jr Physiological studies in congenital heart disease circulatory dynamics in the anomalies of the venous return to the heart including pulmonary arteriovenous fistula Bull Johns Hopkins Hosp 86 0-57 1950
11 Sisson J H G E Murphy and E V Newman Multiple congenital arteriovenous aneurysms in the pulmonary circulation Bull Johns Hopkins Hosp 76 93-111 1945
12 Ferrant M Methemoglobinemia 2 cases in newborn infants caused by nitrates in well water J Pediat. 29 585-592 1946
13 Donahoe W E Cyanosis in infants with nitrates in drinking water as cause Pediatrics 3 308-311 1949
14 Cooke R E Personal communication

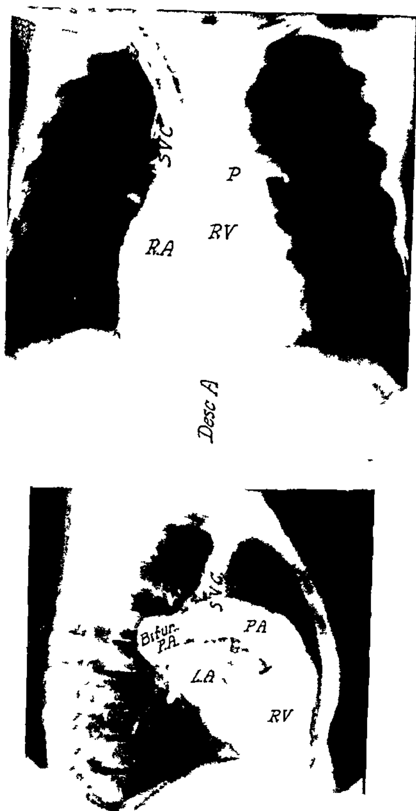


FIGURE XVI-4 Anomalous communication between the right pulmonary artery and the left auricle (Case XVI-2) Child

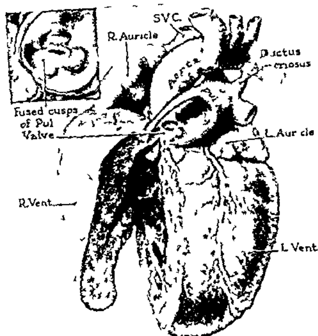


FIGURE XVII–1 Valvular pulmonary stenosis with an intact ventricular septum

poststenotic dilatation develops and eventually the pulmonary artery becomes greatly dilated The right ventricle is normally formed, the ventricular septum is intact

Infundibular stenosis is the name given to the stenosis which involves the infundibular portion of the right ventricle The malformation is caused by the failure in the expansion of the bulbus cordis[2] to form the outflow tract of the right ventricle in the normal manner The outflow tract of the right ventricle may be represented by a narrow pathway which leads to the pulmonary orifice or the infundibular chamber may be separated from the right ventricle by a muscular ridge in which there is only a small opening, as shown in Figure XVII–2 Although stenosis of the infundibular region is the rule in a tetralogy of Fallot its occurrence with an intact ventricular septum is the great exception

The narrowing of the pulmonary orifice, whether it is valvular or infundibular renders it difficult for the right ventricle to pump the blood into the pulmonary artery Consequently the work of the right ventricle is greatly increased and the wall of the right ventricle becomes hypertrophied The smaller the opening into the pulmonary artery, the greater is the difficulty in the ejection of

CHAPTER XVII

PULMONARY STENOSIS

OBSTRUCTION to the pulmonary blood flow is usually caused by an abnormality of the pulmonary valve, in some instances, however, the obstruction occurs within the right ventricle. Both conditions are classified as "pure" pulmonary stenosis because the sole abnormality is the pulmonary stenosis, be it valvular or infundibular (see Section A). Occasionally constrictions occur in the branches of the pulmonary artery (see Section B). The last mentioned condition is designated as "peripheral" pulmonary stenosis.

A. *Pure Pulmonary Stenosis*

"Pure" pulmonary stenosis with an intact ventricular septum is not a rare malformation. With the advent of cardiac catheterization and Sir Russel Brock's operation[1] for the alleviation of pulmonary stenosis, an extraordinary number of cases have been reported. Furthermore, there are all grades of pulmonary stenosis.

NATURE OF THE MALFORMATION

The stenosis is usually of the valvular type, in rare instances it may be located in the infundibulum of the right ventricle. The degree of pulmonary stenosis is subject to great variation. In some instances the pulmonary stenosis is so slight that the condition never causes symptoms and is compatible with a long and active life. In other instances the pulmonary stenosis is so severe that the infant lives only a few months. It has been the author's experience that the pulmonary orifice must be reduced to less than one third of its normal diameter for the condition to cause symptoms. The opening may be only a millimeter or two in diameter. In such instances the infant will die at an early age unless the stenosis is relieved by operation.

The stenosis of the pulmonary valve results from a fusion of the three semilunar cusps over the pulmonary orifice to form a dome which has a central perforation, as shown in Figure XVII–1. Such is the location of the obstruction in approximately 95 per cent of the patients in whom pulmonary stenosis occurs with an intact ventricular septum.

The pulmonary ring is almost always of normal size. The pulmonary artery beyond the valve is usually normal at birth. Over a period of years, however,

As the work of the right ventricle increases, the right ventricle fails to empty completely and the diastolic pressure in that chamber rises With the increase in the diastolic pressure, the right auricle encounters difficulty in emptying itself, consequently the work of the right auricle is increased The readily distensible right auricle undergoes both dilatation and hypertrophy

The valve covering the foramen ovale remains patent in approximately 75 per cent of patients with this malformation When this occurs, the high pressure in the right auricle will eventually push the valve away from the auricular septum and thereby cause functional patency of the foramen ovale Under such circumstances the foramen ovale acts as an escape valve for the relief of the high pressure in the right auricle and at the same time a right to-left shunt is established through it This in turn reduces the volume of blood which is directed into the right ventricle, consequently the right ventricular chamber remains relatively small but the wall of the right ventricle may be enormously thickened it may be more than 1 cm in thickness

COURSE OF THE CIRCULATION

During fetal life inasmuch as little blood flows to the lungs, even a severe degree of pulmonary stenosis places little strain upon the heart Although the increased pressure on the right side of the heart causes more than the usual quota of blood to flow through the foramen ovale to the left auricle and the left ventricle and thereby slightly increases the work of the left ventricle, the difference is not great At birth the heart is approximately normal in size

After birth the increased work demanded of the right ventricle by pulmonary stenosis increases the pressure in the right ventricle and, if this is great, it is transmitted back to the right auricle The increased pressure in that chamber tends to hold the foramen ovale open Therefore, if pulmonary stenosis is extreme the foramen ovale frequently remains widely patent

When the foramen ovale remains open, part of the blood from the right auricle flows into the right ventricle and some blood from the right auricle is shunted into the left auricle The blood which flows into the right ventricle is pumped out through the stenosed pulmonary orifice into the pulmonary artery, thence it flows slowly to the lungs The blood which reaches the lungs takes up its normal quota of oxygen The fully oxygenated blood is returned to the left auricle where it becomes mixed with the venous blood shunted from the right auricle into that chamber This mixture of venous and arterial blood flows from the left auricle into the left ventricle and thence is pumped out through the aorta

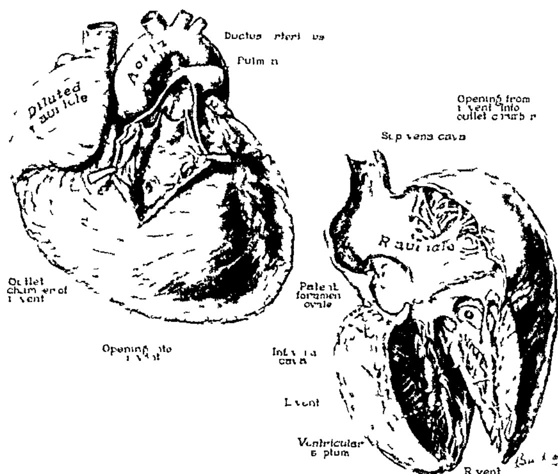


FIGURE XVII-2 Infundibular pulmonary stenosis with an intact ventricular septum (same patient as in Figure XVII-10) Adult

blood from the right ventricle, and the greater the work demanded of that chamber. As the wall of the right ventricle hypertrophies, the musculature at the base of the pulmonary artery becomes thickened and eventually the muscular ring becomes so thick that it actually obstructs the outflow of blood from that chamber. Consequently a vicious circle is set up which leads to still greater hypertrophy.

Furthermore, it seems probable that the fibrotic valve grows more slowly than does the normal valve and therefore the stenosis of the pulmonary orifice becomes proportionally greater with the growth of the individual. Certain it is that this malformation is one which may lead to progressive cardiac enlargement. Since there is no left-to-right shunt, there is no increase in the volume of blood which the right ventricle must pump. Consequently there is little or no dilatation of the right ventricle. The enlargement is almost entirely due to hypertrophy of the muscular wall.

CLINICAL FINDINGS

The clinical findings[38] vary with the severity of the pulmonary stenosis and with the structure of the foramen ovale The more extreme the pulmonary stenosis, the greater is the tendency for the foramen ovale to remain patent Nevertheless, the foramen ovale may become completely sealed even though the pulmonary stenosis is extremely severe

If the pulmonary stenosis is slight, the patient is entirely asymptomatic It is only the presence of a harsh systolic murmur and a weak second sound which indicates that the murmur is of more than functional origin If, on the other hand the stenosis is *severe, the condition* causes progressive cardiac enlargement and, unless the obstruction is relieved, it eventually leads to cardiac failure

The appearance of the patient is remarkable only in that he is usually strong and well developed Infants are often exceptionally sturdy, growth and development are normal

Dyspnea is the outstanding complaint The dyspnea is related to the patient's inability to increase the pulmonary blood flow rather than to a venous-arterial shunt If the pulmonary stenosis is extreme, the patient simply cannot appreciably increase the pulmonary blood flow with exercise This means that he is unable to increase the minute output of the heart and consequently cannot increase the supply of oxygen to the body or to the brain Therefore, even though the patient shows no cyanosis and he has a normal arterial oxygen saturation, he becomes dyspneic on slight exertion and stops to rest Infants with this type of pulmonary stenosis rarely suffer from attacks of paroxysmal dyspnea, children seldom squat when tired

It is important to remember that complaints are late manifestations of this malformation Many patients are asymptomatic until early adult life This fact is vividly illustrated by two patients whom the author has seen One was a man who had served in the Merchant Marine during World War II and at twenty two years of age developed severe cardiac failure The other had been a top athlete in his college class but at twenty four years of age was so incapacitated that he could scarcely walk across the room Indeed, because of his history of athletic prowess and his strong physique combined with the fact that he spent most of the time sitting indolently, one of the author's assistants raised the question whether he could be malingering The enormous cardiac enlargement and chronic cardiac failure clearly indicated that his complaints were real Two days later he keeled over in bed and was dead Fortunately, with the advent of cardiac surgery, this sequence of events is seldom, if ever, seen

to the systemic circulation and is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again (see Diagram xvii–1) It is the establishment of a right to-left shunt through the foramen ovale which renders the patient cyanotic

Since the venous arterial shunt in this malformation is established through the foreman ovale, as long as the foramen ovale is functionally closed and in all instances in which the foramen ovale becomes completely sealed, the course of the circulation is normal The blood from the right auricle flows into the right ventricle and is pumped out with difficulty through the pulmonary orifice into the pulmonary artery Inasmuch as the pulmonary pressure is low, the blood in the pulmonary artery flows slowly to the lungs Nevertheless, all the blood which goes to the lungs is fully oxygenated and is returned in the normal fashion by the pulmonary veins to the left auricle and the left ventricle From the left ventricle the blood is pumped out by way of the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle, there the cycle starts again, as shown in Diagram xvii–2

PHYSIOLOGY OF THE MALFORMATION

The pulmonary stenosis causes difficulty in the expulsion of blood from the right ventricle but it also protects the lungs from the high pressure generated in that chamber Consequently the difficulty imposed upon the circulation is primarily that of the increased work required of the right ventricle If the pulmonary stenosis is extreme, the pulmonary blood flow is low and the systemic blood flow is correspondingly low Nevertheless, if the foramen ovale is sealed, the oxygen saturation of the arterial blood is normal The slow circulation enables the body to take up a large amount of oxygen and nutrition from the blood as it passes through the capillaries The nutrition of the individual is correspondingly good

The stenosis of the pulmonary valve not only increases the work required of the right ventricle to pump the blood into the pulmonary artery, but it also breaks the force with which the blood is ejected into the pulmonary artery, and, furthermore, that force is dissipated in all directions Blood pools in the main pulmonary artery and flows slowly to the lungs The dissipation of the energy generated in the right ventricle is the most probable explanation of the post stenotic dilatation of the pulmonary artery Be that as it may, poststenotic dilatation of the pulmonary artery is the rule In most instances the longer the duration of the pulmonary stenosis and the greater its severity, the greater is the post stenotic dilatation of the pulmonary artery

Diagram XVII–1

Valvular pulmonary stenosis with an intact ventricular septum and a patent foramen ovale

The essential feature of this malformation is the stenosis of the pulmonary valve combined with patency of the foramen ovale The *ductus arteriosus* is completely obliterated

The blood from the right auricle flows both into the right ventricle and through the foramen ovale into the left auricle. The blood from the right ventricle is forced through the stenosed pulmonary orifice to the lungs where it is oxygenated, and the oxygenated blood is returned by the pulmonary veins to the left auricle, where it meets the venous blood which has been forced from the right auricle through the foramen ovale into the left auricle the admixture of venous and arterial blood flows into the left ventricle and out by way of the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle There the cycle starts again

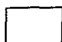
The obstruction to the expulsion of blood from the right ventricle causes dilatation and hypertrophy of that chamber The increased pressure in the right ventricle is transmitted back into the right auricle which becomes enlarged The patency of the foramen ovale acts as an escape valve consequently there is less hypertrophy and dilatation of the right auricle under these conditions than when the foramen ovale is completely closed Moreover there is less engorgement of the liver and it seldom pulsates

Inasmuch as there is high pressure on the right side of the heart and patency of the foramen ovale a right to-left shunt is established between the two auricles The extent of the oxygen unsaturation of the arterial blood depends upon the volume of venous blood shunted from the right auricle to the left auricle As this increases the patient develops cyanosis and polycythemia

Clinical diagnosis The intensity of the cyanosis varies with the severity of the pulmonary stenosis and the extent of patency of the foramen ovale When the stenosis is extreme cyanosis may date from birth Usually however cyanosis appears between the second and the sixth year of life and slowly increases in intensity Dyspnea is more marked than cyanosis Examination of the heart shows a basal systolic murmur and a thrill The second sound at the base is weak or absent There is progressive enlargement of the right ventricle and the right auricle There is fullness of the pulmonary conus which also becomes progressively more marked because of the poststenotic dilatation of the pulmonary artery The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy Terminally the heart becomes enormously enlarged and there is engorgement of the liver and there may be both edema and ascites The condition should however be corrected by surgery before the heart becomes greatly enlarged

Diagram XVII–1

Head and upper extremities
R Lung
L Lung
S.V.C.
P.A.
P.V.
Aorta
Pul. A.
Pul. V.
Pulmonary Artery
L.A.
R.A.
L.V.
R.V.
Descending
Aorta
I.V.C.
Trunk and lower extremities



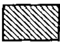
Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

Diagram XVII–2

Valvular pulmonary stenosis with an intact ventricular septum

The sole abnormality in this malformation is the stenosis at the orifice of the pulmonary artery The three semilunar valves are fused together to form a dome with a central perforation The ductus arteriosus is completely obliterated, the foramen ovale is closed

The blood from the right auricle flows into the right ventricle and that which can be *forced* through the pulmonary orifice goes to the lungs and is returned by the pulmonary veins to the left auricle Thence it flows to the left ventricle and out by way of the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle

The obstruction to the outflow of blood from the right ventricle causes dilatation and hypertrophy of that chamber The increased work required of the right ventricle raises the pressure in that chamber and this in turn increases the work required of the right auricle it too is dilated and hypertrophied The inferior vena cava is dilated and the liver is engorged and pulsates

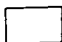
Inasmuch as there is no abnormal communication between the two circulations there is no venous arterial shunt Consequently, the oxygen saturation of the arterial blood is normal

Clinical diagnosis Cyanosis is absent Dyspnea may be extreme There is a basal systolic murmur and a thrill The second sound at the base is weak or absent The heart undergoes progressive enlargement The enlargement is mainly due to hypertrophy of the right ventricle and to dilatation and hypertrophy of the right auricle There is fullness of the pulmonary conus due to poststenotic dilatation of the main pulmonary artery The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy Terminally the liver is engorged and pulsates and there may be both edema of the extremities and ascites

The condition can and should be corrected by surgery as soon as there is evidence of cardiac strain

DIAGRAM XVII–2

Head and upper extremities
S.V.C.
P.A.
R Lung
PV
Aorta
Pul. A.
L Lung
Pul V
Pulmonary Artery
L.A
Descending
R.A.
LV
RV
I.V.C.
Aorta
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

are more frequently palpable with a closed auricular septum or with slight patency of the foramen ovale than when the valve is widely patent Regardless of the presence or absence of cyanosis, pulsations of the liver are indicative of high pressure in the right auricle and hence of a severe degree of pulmonary stenosis Therefore this finding is a strong indication for prompt operation

Edema and ascites may occur as late manifestations of the right sided heart failure Nevertheless, owing to the difficulty in the expulsion of blood from the right ventricle, pulmonary congestion is rare

Episodes of collapse and loss of consciousness are not uncommon terminal manifestations, because the patient suffers from a low pulmonary blood flow and a correspondingly low systemic blood flow Indeed, there may be a striking discrepancy between the patient s sturdy growth and his history of former activity and the sudden development of lassitude and exhaustion As a matter of fact the pulmonary blood flow may become so meager that life just peters out

CARDIAC FINDINGS

The size of the heart and the rate of enlargement vary with the severity of the pulmonary stenosis If the pulmonary stenosis is slight to moderate, the malformation does not cause cardiac enlargement When the pulmonary stenosis is severe, the condition leads to progressive enlargement *Cardiac enlargement precedes symptoms* Therefore it is extremely important for patients suspected of this malformation to be kept under close observation This is especially true for infants as cardiac enlargement may occur with extraordinary rapidity Moreover, months are to an infant as years are to a child of ten Therefore the infant should be followed at monthly intervals for evidence of increase in heart size The two x rays in Figure XVII–3 show a tremendous increase in the size of the heart This occurred in an infant between six months and one year of age If the heart becomes definitely enlarged or the infant develops pulsations at the margin of the liver or shows signs of cardiac failure, prompt operation is indicated

In children however cardiac enlargement occurs more slowly For this reason changes in the electrocardiogram are a more sensitive index to increasing cardiac strain than are the changes in the x ray (see Electrocardiographic Findings) Cardiac enlargement may also occur rapidly during the spurt of growth which occurs at puberty Therefore at this age, changes in the size of heart again become significant

The rate of enlargement depends upon the amount of work required of the

The occurrence of cyanosis depends primarily upon the structure of the auricular septum[4] If the auricular septum is intact and the foramen ovale is completely sealed, there is no communication between the two sides of the heart Even though the pressure in the right auricle is greatly increased, there is no venous arterial shunt Cyanosis is absent The oxygen saturation of the arterial blood is normal

Since the foramen ovale is normally patent at birth, in the presence of pulmonary stenosis a right to-left shunt may be readily established immediately after birth For this reason infants with pulmonary stenosis frequently show cyanosis at birth Generally, however, the foramen ovale tends to close, the right to-left shunt lessens, and not infrequently cyanosis entirely disappears

On the other hand, if the foramen ovale remains patent, as the pulmonary stenosis increases the pressure in the right ventricle rises and this in turn increases the pressure in the right auricle, under such circumstances the foramen ovale is again forced open and the right to-left shunt is established When the volume of the shunt is sufficiently great, the patient develops cyanosis Consequently, although cyanosis due to patency of the foramen ovale may date from birth, or may be present at birth and subsequently disappear, it frequently develops insidiously and becomes apparent when the patient is between two and seven years of age Thereafter, unless the stenosis is relieved, the cyanosis gradually increases in intensity

Polycythemia develops as the oxygen unsaturation of the arterial blood increases The red blood cell count, the amount of available hemoglobin, and the hematocrit reading may reach as high levels in these patients as in any other patient who has a persistent venous arterial shunt The increase in the hemoglobin in turn increases the intensity of the cyanosis

Clubbing of the extremities gradually develops as the polycythemia increases

The venous pressure becomes increased when the pressure in the right auricle becomes abnormally high

The neck vessels become engorged and often show conspicuous pulsations

The liver gradually enlarges and frequently extends from two to three finger breadths below the costal margin Owing to the high pressure on the right side of the heart and the difficulty in the expulsion of blood from the right auricle, the forceful contractions of the right auricle are frequently transmitted back to the margin of the liver Such pulsations are presystolic in time and may occur with minimal engorgement of the liver Inasmuch as the foramen ovale acts as an escape valve for the high pressure in the right auricle, pulsations in the liver

are more frequently palpable with a closed auricular septum or with slight patency of the foramen ovale than when the valve is widely patent Regardless of the presence or absence of cyanosis, pulsations of the liver are indicative of high pressure in the right auricle and hence of a severe degree of pulmonary stenosis Therefore this finding is a strong indication for prompt operation

Edema and ascites may occur as late manifestations of the right sided heart failure Nevertheless, owing to the difficulty in the expulsion of blood from the right ventricle, pulmonary congestion is rare

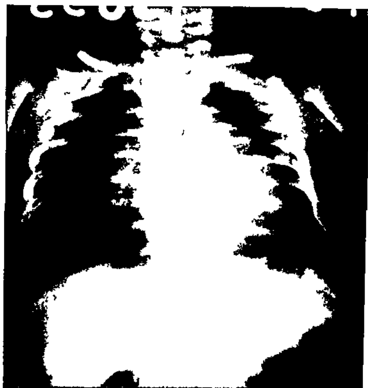
Episodes of collapse and loss of consciousness are not uncommon terminal manifestations, because the patient suffers from a low pulmonary blood flow and a correspondingly low systemic blood flow Indeed, there may be a striking discrepancy between the patient s sturdy growth and his history of former activity and the sudden development of lassitude and exhaustion As a matter of fact the pulmonary blood flow may become so meager that life just peters out

CARDIAC FINDINGS

The size of the heart and the rate of enlargement vary with the severity of the pulmonary stenosis If the pulmonary stenosis is slight to moderate, the malformation does not cause cardiac enlargement When the pulmonary stenosis is severe the condition leads to progressive enlargement *Cardiac enlargement precedes symptoms* Therefore it is extremely important for patients suspected of this malformation to be kept under close observation This is especially true for infants as cardiac enlargement may occur with extraordinary rapidity Moreover, months are to an infant as years are to a child of ten Therefore the infant should be followed at monthly intervals for evidence of increase in heart size The two x rays in Figure XVII–3 show a tremendous increase in the size of the heart This occurred in an infant between six months and one year of age If the heart becomes definitely enlarged, or the infant develops pulsations at the margin of the liver or shows signs of cardiac failure, prompt operation is indicated

In children however cardiac enlargement occurs more slowly For this reason changes in the electrocardiogram are a more sensitive index to increasing cardiac strain than are the changes in the x ray (see Electrocardiographic Findings) Cardiac enlargement may also occur rapidly during the spurt of growth which occurs at puberty Therefore at this age, changes in the size of heart again become significant

The rate of enlargement depends upon the amount of work required of the



At six months



At one year

FIGURE XVII–3 Severe valvular pulmonary stenosis with an intact ventricular septum Infant

right ventricle Therefore the size of the heart varies not only with the severity of the pulmonary stenosis, but also with the load placed on the heart by physical exertion The author has seen one woman with severe pulmonary stenosis and a pressure of nearly 200 mm of mercury in the right ventricle who had lead such a quiet, restricted life that her heart had remained normal in size Usually severe pulmonary stenosis leads to great cardiac enlargement

Rapid increase in the size of the heart during childhood at the period when growth is slow should suggest the possibility that the stenosis is *infundibular*, not valvular, in type

A harsh systolic murmur and a thrill are produced as the blood is forced through the stenosed pulmonary orifice into the pulmonary artery Both the murmur and the thrill are maximal over the pulmonary area The thrill is often readily palpable in the suprasternal fossa In cases of severe pulmonary stenosis and great cardiac enlargement, the murmur is well heard high up beneath the left clavicle and is frequently well transmitted to the interscapular region The fibrosis of the valve, combined with the low pressure in the pulmonary artery, prevents an abrupt or snapping closure of this valve, consequently the pulmonic s cond sound is weak or absent. Nevertheless, the murmur ends abruptly with the end of systole The pulmonary valve is usually competent, diastole is clear It is the harsh systolic murmur that ends abruptly, combined with a weak or absent second sound, which gives the clue to the diagnosis

It is important to remember that an infant may have such an extreme degree of pulmonary stenosis that there is no murmur or thrill, such an infant suffers from rapid cardiac enlargement and cardiac failure associated with an engorged pulsating liver

In the presence of great cardiac enlargement there is usually an apical systolic murmur which is transmitted to the axilla and also a gallop rhythm

The characteristic thrill in the suprasternal notch is absent when there is infundibular stenosis even when the murmur and thrill are maximal high up in the second interspace to the left of the sternum The murmur and the thrill are frequently of maximal intensity at a lower level in infundibular stenosis than in valvular stenosis of a comparable degree Occasionally the second sound is audible over the base of the heart, this finding is also suggestive of an infundibular stenosis

Cardiac failure may occur in the neonatal period without great cardiac enlargement It is manifested by cyanosis and by engorgement of the liver with pulsations at its margin There may even be generalized edema The infant may

be greatly helped by digitalis As the baby regains compensation, cyanosis lessens and may disappear and thereafter he may do well for a number of months or even for years

In contrast to this, if the infant does well in the neonatal period but during the subsequent months develops cardiac enlargement and cardiac failure, early operation is urgently indicated (see under Treatment)

In children and adults cardiac failure is a late manifestation As long as the heart is able to enlarge to meet the needs of the individual, the patient remains asymptomatic Consequently, by the time cardiac failure develops, the heart is always greatly enlarged The liver is engorged and usually pulsations are palpable at its margin Edema develops and subsequently there may be ascites The lungs remain relatively clear Even at this time prompt relief of the pulmonary stenosis may be of great benefit to the patient

X RAY AND FLUOROSCOPIC FINDINGS

The x ray and fluoroscopic findings are distinctive The second curve to the left of the sternum is exaggerated If the stenosis is slight or moderate, the heart may be normal in size Usually the right ventricle is hypertrophied and the heart appears to be slightly to moderately enlarged Owing to the development of poststenotic dilatation of the pulmonary artery, in children and adults the main pulmonary artery is usually enlarged and there is fullness in the region of the pulmonary conus, as shown in Figures xvii–4, 5, 6, and 15

When the pulmonary stenosis is extreme, the heart may become enormously enlarged The enlargement of the right ventricle is so great that it not only presses against the anterior chest wall but also displaces the left ventricle backward and the pulmonary artery upward The greatly dilated pulmonary artery lies above the thick walled right ventricle and the pulmonary valve and the main segment of the pulmonary artery lie at an abnormally high level

The absence of poststenotic dilatation of the pulmonary artery is sometimes considered indicative of infundibular stenosis There are, however, many exceptions The characteristic fullness of the pulmonary conus is frequently absent in early infancy, as poststenotic dilatation takes time to develop Indeed, in children with extreme pulmonary stenosis (see Figure xvii–8), so little blood may be pumped through the pulmonary orifice that poststenotic dilatation of the pulmonary artery does not occur Furthermore, the characteristic fullness of the pulmonary conus may occur in children with infundibular stenosis as illustrated in Figure xvii–9 Nevertheless, in older patients with great cardiac enlargement, the

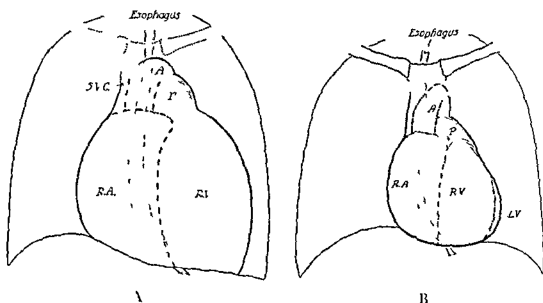


Anterior posterior position

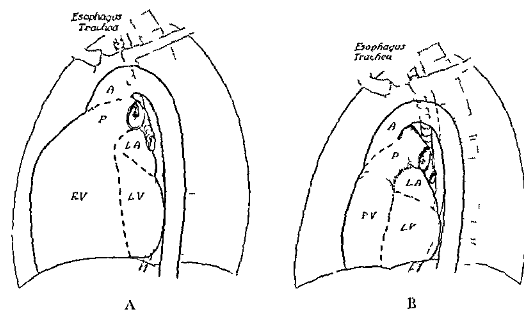


Left anterior oblique position

FIGURE XVII-4 Valvular pulmonary stenosis with an intact ventricular septum (same patient as in Figures XVII-13 14) Child



ANTERIOR POSTERIOR POSITION



LEFT ANTERIOR OBLIQUE POSITION

FIGURE XVII-5 (A) Valvular pulmonary stenosis with an intact ventricular septum and (B) normal heart Child



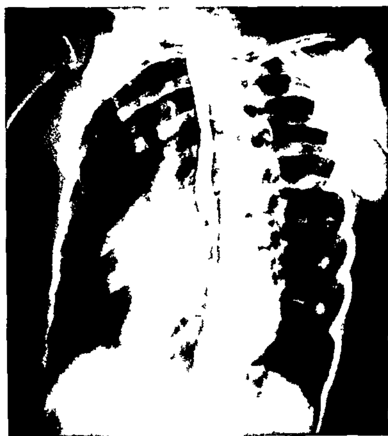
FIGURE XVII-6 Valvular pulmonary stenosis with an intact ventricular septum (same patient as in Figure XVII-7) Adult

absence of fullness of the pulmonary conus, as shown in Figure XVII-10, usually is indicative of infundibular stenosis

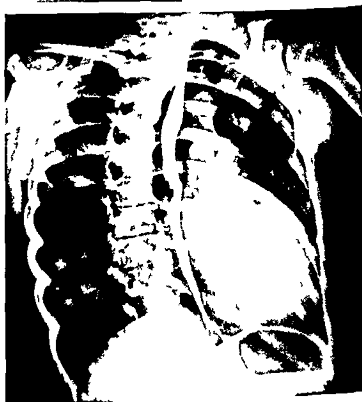
In the left anterior-oblique position the right ventricle is usually seen to be enlarged (see Figures XVII-4, 5, and 7) When the heart is greatly enlarged, the right ventricle becomes flattened against the sternum[4] and further enlargement of the right ventricle displaces the left ventricle backward so that the heart does not clear the spinal column until the patient is rotated to an angle of 60 degrees Under such circumstances, it is usually possible to detect the interventricular groove The posterior position of this groove adds confirmatory evidence that the increase in the size of the heart is mainly due to the great right ventricular hypertrophy (see Figure XVII-5)

Examination in the right anterior-oblique position contributes little, except that the left auricle is of normal size and the esophagram is normal

Fluoroscopic examination aids still further in the diagnosis as there is a striking discrepancy between the size of the pulmonary conus and the pulsations



Left anterior-oblique position



Right anterior oblique position

Figure xvii-7 Valvular pulmonary stenosis with an intact ventricular septum (same patient as in Figure xvii-6) Adult



Before operation



After operation

FIGURE XVII–8 Severe valvular pulmonary stenosis with an intact ventricular septum Child



FIGURE XVII-9 Infundibular pulmonary stenosis with an intact ventricular septum Child



FIGURE XVII-10 Infundibular pulmonary stenosis with an intact ventricular septum (same patient as in Figure XVII-2) Adult

in the main branches of the pulmonary arteries Even if there are visible pulsations in the pulmonary conus, the pulsations in the left and right branches of the pulmonary artery are minimal or absent The greater the cardiac enlargement, the quieter are the hilar shadows In patients with minimal cardiac enlargement, a hilar dance may be discerned, in those with patency of the foramen ovale there is an actual reduction in the pulmonary blood flow and the lungs are proportionately clear

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram is of great aid not only in the diagnosis of this malformation but in an estimation of the severity of the pulmonary stenosis A right axis deviation and evidence of right ventricular hypertrophy are characteristic of "pure" pulmonary stenosis Furthermore, as the work of the right ventricle increases, the unipolar precordial leads show evidence of progressively greater right ventricular hypertrophy and so-called right ventricular "strain"

When the pulmonary stenosis is extremely mild, the electrocardiogram may show only a right axis deviation and the unipolar precordial leads will be normal As the work of the right ventricle increases, the unipolar precordial leads reflect the progressive right ventricular hypertrophy When the pressure is under 80 mm of mercury, the R wave in V_1 is usually prominent but the T wave remains upright As the pressure in the right ventricle increases, the T waves in V_1 become inverted This usually does not occur until the pressure is between 100 and 120 mm of mercury As the pressure in the right ventricle rises still further, the pattern of so-called right ventricular "strain" develops that is, a slight delay in the onset of the intrinsicoid deflection of V_1 and a tall R wave combined with inversion of the T waves in V_1, possibly in V_3 and even in V_5, the T wave in V_6 tends to become upright the S waves over the left precordium are deep Such changes (see Figure XVII–11) are usually indicative of a right ventricular pressure of more than 140 mm of mercury[7]

The changes in the electrocardiogram usually become apparent before there is any detectable enlargement in the x ray Therefore serial electrocardiograms are extremely useful in the evaluation of the condition of the patient Careful evaluation of the electrocardiographic findings eliminates the necessity for cardiac catheterization

In childhood electrocardiographic evidence of rapidly progressive right ventricular strain which is out of proportion to an increase in the size of the heart is always suggestive that the stenosis is infundibular and not valvular

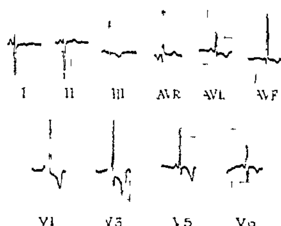


Figure XVII-11 Valvular pulmonary stenosis

The P waves reflect the amount of work required of the auricle As the pressure in the right ventricle rises, the pressure in the right auricle also rises and the right auricle undergoes dilatation and hypertrophy Under such circumstances the P waves are both broad and tall, consequently they resemble the Appalachian Mountains rather than the tall, sharp peaks of the Himalayas

SPECIAL TESTS

The circulation time (arm to tongue) is usually normal or prolonged Even though the foramen ovale is widely patent, sufficient test material is seldom shunted through the foramen ovale to the systemic circulation to give a short circulation time The circulation time is commonly from twelve to fifteen seconds in a child and may be over twenty seconds in an adult In infants the normal circulation time is so short that the test is seldom significant, indeed, in the presence of a widely patent foramen ovale the circulation time may be phenomenally short

The oxygen saturation of the arterial blood varies with the amount of venous blood which is shunted through the foramen ovale If the foramen ovale is widely patent, the oxygen saturation of the arterial blood may be greatly reduced Usually, however, even in the presence of persistent cyanosis, the arterial oxygen saturation is relatively high and falls but slightly with exercise In patients with an intact auricular septum, the oxygen saturation of the arterial blood is normal

The exercise test usually shows no increase in the oxygen consumption per liter of ventilation [8] In other words, the pulmonary stenosis prevents the increase in the pulmonary blood flow which normally occurs during exercise

Cardiac catheterization reveals a high pressure in the right ventricle and a

low pressure in the pulmonary artery, frequently the pressure in the right auricle is also increased The oxygen content of the blood in the right auricle, in the right ventricle, and in the pulmonary artery should be essentially the same Although the over all shunt is from right to left, if the catheter is drawn into the stream of the shunt and the sample is taken close to the opening in the auricular septum the oxygen content of this sample may be higher than that taken from the superior vena cava, thereby giving the impression that there is a gross defect in the auricular septum Occasionally, even though there is no gross defect in the auricular septum, the catheter may be passed through the foramen ovale into the left auricle When the pulmonary stenosis is extreme, it may be difficult or impossible to pass the catheter into the pulmonary artery

If the pressure in the right ventricle exceeds systemic pressure, it is strong presumptive evidence that the ventricular septum is intact It is, however, essential to catheterize the pulmonary artery because only the combination of high pressure in the right ventricle and low pressure in the pulmonary artery proves the existence of pulmonary stenosis Figure XVII-12 shows the abrupt change in the pressure at the level of the pulmonary valve as the catheter is withdrawn from the pulmonary artery into the right ventricle

A few words of caution in regard to catheterization are necessary Serious symptoms may develop if the size of the catheter is such as to occlude the pulmonary orifice and thereby cut off the supply of blood to the lung Furthermore, in the presence of great right ventricular hypertrophy there is real danger that catheterization of the right ventricle may precipitate ventricular fibrillation Finally it should be remembered that with myocardial failure, the pressure in the right ventricle will fall

Cardiac catheterization may be of value in the differentiation of infundibular from valvular stenosis In valvular stenosis the catheter passes directly from an area of high pressure to an area of low pressure When the stenosis lies within the ventricle, the catheter passes from the area of high pressure through an area of lower ventricular pressure before it enters the pulmonary artery Furthermore, when one is unable to pass the catheter into the pulmonary artery, the obstruction is usually encountered at a lower level in an infundibular stenosis than in a stenosis of the valvular type Nevertheless, unless the physician is on his guard, the differences upon catheterization may be overlooked

Angiocardiography shows prompt opacification of the right side of the heart, first the right auricle and then the right ventricle is filled It is usually possible to demonstrate that the right ventricle is thick walled as the dye does not extend

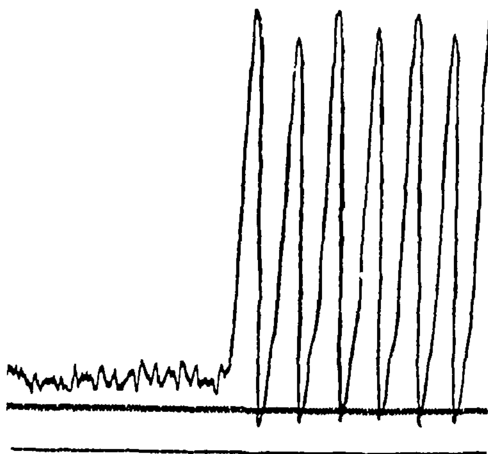


FIGURE XVII-12 Valvular pulmonary stenosis

Pressure tracing showing abrupt change in pressure between the pulmonary artery and the right ventricle Pressure recording pulmonary artery 22/9 mm Hg right ventricle 140/4 mm Hg

to the margin of the cardiac silhouette The pulmonary artery is promptly opacified Nevertheless, owing to the pulmonary stenosis, only a small amount of dye is ejected into the pulmonary artery with each ventricular systole Consequently, although the main pulmonary artery fills immediately, it continues to fill over a long period of time The aorta is not promptly delineated and is never opacified before the left auricle and the left ventricle are filled The latter are always better visualized in the lateral position than in the anterior posterior position Therefore angiocardiograms in both positions are important When there is patency of the foramen ovale due to the constant shunting of a small amount of blood through the foramen ovale, the aorta is usually faintly outlined and dye in small concentration remains in the aorta for several seconds and then disappears In as

much as the blood flows slowly from the pulmonary artery to the lungs, dye in high concentration may remain in the main pulmonary artery after it has disappeared from the aorta Figures XVII-13 and 14 show the lingering of the dye in the pulmonary artery in a child with 'pure' pulmonary stenosis

In contrast to this, the angiocardiogram of an infant with a widely patent foramen ovale may show such prompt and dense opacification of the aorta simultaneously with the delineation of the pulmonary artery that the condition may be mistaken for a tetralogy of Fallot

A word of warning in regard to angiocardiography if the pulmonary stenosis is extreme, throughout the time that the dye is being ejected into the pulmonary artery, the circulation of blood to the lungs is virtually cut off For this reason angiocardiography is dangerous for patients with severe pulmonary stenosis, it is especially dangerous for those with an extremely low arterial oxygen saturation Indeed, the deprivation of oxygen may be fatal

DIAGNOSIS

The outstanding features of this malformation are a harsh systolic murmur and a thrill over the pulmonary area, combined with a weak or absent second sound at the base The patient may or may not show cyanosis The insidious development of cyanosis in childhood is common The demonstration of pulsations at the margin of the liver indicates that the stenosis is extremely severe

The electrocardiogram shows a right axis deviation and generally shows the pattern of right ventricular hypertrophy X ray examination almost invariably shows fullness of the pulmonary conus and enlargement of the main branches of the pulmonary artery Upon fluoroscopy there is a discrepancy between the prominence of the pulmonary conus and the pulsations in the hilar shadows the former is conspicuous the latter are minimal or absent

The diagnosis can be definitively established by cardiac catheterization provided the pulmonary artery is catheterized When there is no evidence of a left to-right intracardiac shunt the finding of high pressure in the right ventricle combined with low pressure in the pulmonary artery is indicative of pulmonary stenosis with an intact ventricular septum

The differentiation of infundibular from valvular stenosis although difficult and indeed sometimes impossible, is extremely important *In children* the evidence of rapid cardiac enlargement in the x ray or of rapidly progressive right ventricular hypertrophy in the electrocardiogram should suggest that the stenosis is infundibular The finding of a murmur maximal in the second left inter

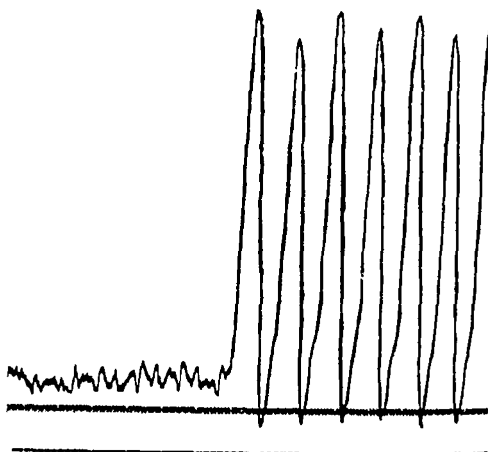


FIGURE XVII-12 Valvular pulmonary stenosis

Pressure tracing showing abrupt change in pressure between the pulmonary artery and the right ventricle Pressure recording pulmonary artery 2[illegible]/9 mm Hg right ventricle 140/4 mm Hg

to the margin of the cardiac silhouette The pulmonary artery is promptly opacified Nevertheless, owing to the pulmonary stenosis, only a small amount of dye is ejected into the pulmonary artery with each ventricular systole Consequently, although the main pulmonary artery fills immediately, it continues to fill over a long period of time The aorta is not promptly delineated and is never opacified before the left auricle and the left ventricle are filled The latter are always better visualized *in the lateral position than in the anterior posterior position* Therefore angiocardiograms in both positions are important When there is patency of the foramen ovale due to the constant shunting of a small amount of blood through the foramen ovale, the aorta is usually faintly outlined and dye in small concentration remains in the aorta for several seconds and then disappears Inas



At four seconds



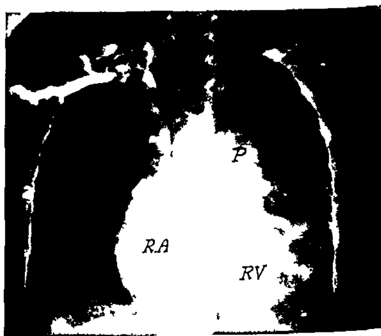
At seven seconds

FIGURE XVII-14 Valvular pulmonary stenosis with an intact ventricular septum (series from Figure XVII-13 continued same patient as in Figure XVII-4) Child

Note dye still visible in the main pulmonary artery at seven seconds



At one second



At two seconds

FIGURE XVII-15 Valvular pulmonary stenosis with an intact ventricular septum (same patient as in Figure XVII-4) Child

Ebstein s anomaly of the tricuspid valve when extreme, may closely resemble severe long standing pulmonary stenosis In Ebstein s anomaly fatigue is the outstanding complaint, cyanosis is more marked than dyspnea, the heart sounds are weak, the murmurs are blurred and confused, and arrhythmias are common Compensation is frequently precarious but pulsations at the margin of the liver are usually absent The electrocardiogram usually differentiates the two conditions in Ebstein s anomaly the deflections in V_1 are of low amplitude and the QRS is of long duration, whereas in pulmonary stenosis the R wave in V_1 is tall Angiocardiography shows enormous dilatation of the right auricle and an extremely thin walled right ventricle Cardiac catheterization shows increased pressure in the right auricle and normal pressure in the right ventricle and in the pulmonary artery

Complete transposition of the great vessels may be confused with pulmonary stenosis during the first weeks of life In both conditions cyanosis may date from birth the heart is normal in size, there is no fullness of the pulmonary conus, and the vascular markings in the lungs are normal or slightly increased Moreover, both conditions may cause cardiac failure at an early age In pure pulmonary stenosis however, the electrocardiogram shows greater evidence of right ventricular hypertrophy than is usual in a complete transposition of the great vessels The clinical course is also different In pure pulmonary stenosis the infant usually responds to digitalis and regains compensation cyanosis gradually diminishes and may entirely disappear Thereafter the infant gains weight and does well In a complete transposition of the great vessels of such a type as to cause cyanosis and cardiac failure in the neonatal period, although the infant may be temporarily helped by digitalis the clinical course is one of increasing cyanosis rapid cardiac enlargement and cardiac failure Death usually occurs within the first few weeks of life

A tetralogy of Fallot may be confused with an isolated valvular pulmonary stenosis in early infancy In a tetralogy of Fallot the lung fields are excessively clear Furthermore the heart is usually normal in size, whereas a severe degree of pulmonary stenosis with an intact ventricular septum leads to great cardiac enlargement A child with a tetralogy of Fallot develops cyanosis at an early age and usually squats when tired the heart is small, the pulmonary artery is inconspicuous the liver does not pulsate and the circulation time is short The electrocardiogram shows right axis deviation and evidence of right ventricular hypertrophy but usually does not show the pattern of extreme right ventricular hypertrophy In doubtful cases cardiac catheterization may be necessary to differ

space and no thrill in the suprasternal notch is also strongly suggestive of in fundibular stenosis *In adults* the absence of demonstrable poststenotic dilatation of the pulmonary artery or the low level at which the systolic murmur is maxi mal is suggestive of an infundibular stenosis

DIFFERENTIAL DIAGNOSIS

In the absence of cyanosis "pure' pulmonary stenosis may be confused with an auricular septal defect or with pulmonary hypertension Both the non-cy anotic and the cyanotic forms may be mistaken for an Eisenmenger complex and occasionally may be confused with Ebstein's anomaly of the tricuspid valve In early infancy the cyanotic form may be mistaken for a complete transposition of the great vessels, both in infancy and in childhood the condition may occa sionally be mistaken for a tetralogy of Fallot and for defective development of the right ventricle with pulmonary stenosis

An auricular septal defect is differentiated from 'pure" pulmonary stenosis by a slightly accentuated second sound at the base of the heart, which is usually reduplicated Furthermore, fluoroscopy reveals the presence of a hilar dance The electrocardiogram usually shows a right bundle branch block Furthermore, the patient has a frail build but does not suffer from severe dyspnea In pure pulmonary stenosis the second heart sound is weak or absent and, although the pulmonary artery may be conspicuous, the pulsations in its branches are mini mal The unipolar precordial leads show evidence of right ventricular hyper trophy The patient, although of sturdy stature, may become dyspneic on slight exertion

Pulmonary hypertension is differentiated by the fact that the pulmonic sec ond sound is markedly accentuated and is usually reduplicated Indeed, it is only in the presence of myocardial failure that the second sound may become so weak that the two conditions are confused Furthermore, pulmonary insufficiency is relatively common with pulmonary hypertension and rarely occurs with pul monary stenosis

The Eisenmenger complex is differentiated from pure' pulmonary stenosis by the absence of cyanosis and dyspnea in infancy and early childhood, by the presence of a hilar dance, and by the fact that the circulation time is short In older patients angiocardiography will show clear evidence of an over riding aorta Cardiac catheterization will show that the oxygen saturation in the pul monary artery is higher than that in the right ventricle and that the pressure in the pulmonary artery is abnormally high

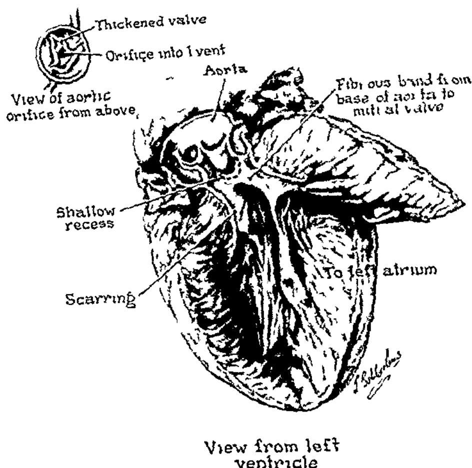


FIGURE XVII–16 Valvular pulmonary stenosis combined with subvalvular aortic stenosis (same patient as in Figures XVII–17 18) Child

valvular pulmonary stenosis an intact ventricular septum, and a gross defect in the auricular septum

Pulmonary stenosis may occur in combination with aortic stenosis[9, 10] The author has seen one such instance in a five year old child who proved to have a valvular pulmonary stenosis and subvalvular aortic stenosis (see Figure XVII–16) This case was reported by Sissman et al[11] The patient had a harsh systolic murmur maximal over the base of the heart, which obliterated the heart sounds The second sound was inaudible to the right as well as to the left of the sternum The child suffered from progressive cardiac enlargement and cardiac failure The



FIGURE XVII–15 Valvular pulmonary stenosis an auricular septal defect, and an intact ventricular septum Adult

entiate the two conditions If the right ventricular pressure is above systemic pressure, it is strong presumptive evidence that the ventricular septum is intact

Defective development of the right ventricle with pulmonary stenosis differs from 'pure pulmonary stenosis in that the right ventricle is a small chamber which is unable to carry its full load Because of the decreased pulmonary blood flow and the small size of the pulmonary artery, the shadow at the base of the heart to the left of the sternum is concave In the left anterior oblique position the x ray shows that the left ventricle as well as the right ventricle is enlarged The electrocardiogram shows evidence of right ventricular hypertrophy in V_1 and left ventricular dominance in V and V_6 Angiocardiography shows that the right ventricle is a tiny chamber

COMMONLY ASSOCIATED ANOMALIES

A gross defect in the auricular septum of the ostium secundum type may occur in combination with valvular pulmonary stenosis Under such circumstances the defect in the auricular septum acts as an escape valve Consequently the more severe the pulmonary stenosis, the greater is the right to left shunt and the deeper is the cyanosis The patient may be severely limited but the heart remains remarkably small Figure XVII–15 illustrates the case of an adult with a severe

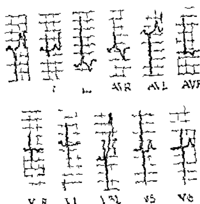


FIGURE XVII-18 Valvular pulmonary stenosis combined with subvalvular aortic stenosis (same patient as in Figure XVII-16) Child

Prophylactic antibiotics are indicated throughout life for the prevention of subacute bacterial endocarditis when there is danger of infection from dental extractions or other procedures Even after successful operation, the pulmonary valve is not normal the patient is still susceptible to the disease

Limitation of exercise is rarely necessary, as the condition can be relieved by surgery Therefore only if surgical treatment is unavailable is restriction of activity indicated In most parts of the world a child with a valvular pulmonary stenosis may be permitted to lead a normal life, if his activity places a strain on his heart, operation is indicated

The advisability of surgical treatment depends upon the severity of the stenosis In some instances the pulmonary stenosis is so slight that the heart is normal in size, the pulmonary blood flow can be increased with exercise, and the electrocardiogram does not even show evidence of right ventricular hypertrophy Such a patient is asymptomatic and if he has attained his full growth, he will certainly remain so The condition need cause no concern The prognosis is excellent without operation

In the author s opinion, if the heart remains normal in size and the patient is asymptomatic and able to lead a normal life and the pressure is under 100 mm of mercury in the right ventricle, operation *is not indicated*, even if the electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy It must be remembered that operation *does not* restore the valve to normal and that mild pulmonary stenosis is compatible with a long and active life

Surgical treatment is indicated if there is evidence of *severe* or *progressive* cardiac strain Inasmuch as cardiac *enlargement precedes* symptoms, the severity of the condition must be judged on *objective* findings During infancy the best



FIGURE XVII-17 Valvular pulmonary stenosis combined with subvalvular aortic stenosis (same patient as in Figure XVII-16) Child

pulses were of good volume The blood pressure was normal The x ray showed great cardiac enlargement with a slightly upturned rounded apex, the pulmonary vascularity was normal or slightly reduced (see Figure XVII-17) The electrocardiogram showed a right axis deviation and evidence of right ventricular hypertrophy It differed from the electrocardiogram usually seen in severe pulmonary stenosis in that V1 showed deep S waves in addition to conspicuous R waves, and V3 showed unusually high T waves In this instance the electrocardiographic findings were the only ones which offered a clue to the existence of the aortic lesion (see Figure XVII-18)

TREATMENT

Medical treatment is indicated in an infant who develops cardiac failure in the neonatal period The infant should rapidly receive his calculated dose of digitalis or one of the allied preparations With such treatment he usually regains compensation and thereafter does well for many months and often for years *If he fails to respond to digitalis, immediate operation is indicated*

Furthermore, if the infant does well in the neonatal period and subsequently develops cardiac failure with a large pulsating liver, prompt operation is indicated Indeed, the condition constitutes a veritable surgical emergency

Digitalis, except in the neonatal period, is seldom indicated If, however, the patient shows signs of cardiac failure, digitalis and diuretics are of value Such treatment is purely palliative, prompt operation is imperative

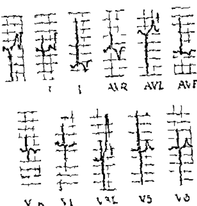


FIGURE XVII-18 Valvular pulmonary stenosis combined with subvalvular aortic stenosis (same patient as in Figure XVII-16) Child

Prophylactic antibiotics are indicated throughout life for the prevention of subacute bacterial endocarditis when there is danger of infection from dental extractions or other procedures Even after successful operation, the pulmonary valve is not normal the patient is still susceptible to the disease

Limitation of exercise is rarely necessary, as the condition can be relieved by surgery Therefore only if surgical treatment is unavailable is restriction of activity indicated In most parts of the world a child with a valvular pulmonary stenosis may be permitted to lead a normal life, if his activity places a strain on his heart, operation is indicated

The advisability of surgical treatment depends upon the severity of the stenosis In some instances the pulmonary stenosis is so slight that the heart is normal in size, the pulmonary blood flow can be increased with exercise, and the electrocardiogram does not even show evidence of right ventricular hypertrophy Such a patient is asymptomatic and, if he has attained his full growth, he will certainly remain so The condition need cause *no concern* The prognosis is excellent without operation

In the author s opinion if the heart remains normal in size and the patient is asymptomatic and able to lead a normal life and the pressure is under 100 mm of mercury in the right ventricle operation is not indicated, even if the electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy It must be remembered that operation does *not* restore the valve to normal and that mild pulmonary stenosis is compatible with a long and active life

Surgical treatment is indicated if there is evidence of severe or progressive cardiac strain Inasmuch as cardiac enlargement precedes symptoms, the severity of the condition must be judged on objective findings During infancy the best

guide is the size of the heart in the x ray If it is but slightly enlarged, serial x rays at monthly or bimonthly intervals will reveal whether or not the condition is progressive During childhood enlargement occurs more slowly, therefore the electrocardiogram offers the best index of the amount of work required of the right ventricle Regardless of the electrocardiogram, cyanosis which persists in childhood is always an indication for operation as it occurs only when the pressure in the right auricle is greater than that in the left auricle

In brief, *the indications for operation* are persistent cyanosis, or a right ventricular pressure of over 100 mm of mercury combined with evidence of right ventricular hypertrophy in the electrocardiogram or cardiac enlargement The ideal time for operation depends not on the age of the child, but on the size of the heart It is wise to operate prior to the occurrence of great cardiac enlargement or cardiac failure Therefore, if there is progressive increase in the size of the heart out of proportion to the growth of the patient, operation is indicated In young infants immediate operation is indicated if there is great cardiac enlargement or pulsations palpable at the margin of the liver

Several types of operation have been developed The first was that of valvulotomy developed by Sir Russel Brock,[1] who used the transventricular approach to cut the pulmonary valve and dilate the pulmonary orifice This operation has been simplified by Potts[12] by the invention of special instruments Swan and his associates[13] have advocated the use of hypothermia, so that the valve may be approached from the pulmonary artery and divided under direct vision Søndergaard[14] has developed a technique by which the operation may be accomplished through the pulmonary artery but without hypothermia Recently a number of surgeons have advocated operation under direct vision with the aid of a pump and oxygenator

At operation the surgeon is usually able to confirm the diagnosis by the finding of a large pulmonary artery in which the pressure is low Frequently, with each ventricular systole, he can feel the forceful thrust of the pulmonary valve against his finger, or he may feel the jet of blood forced through the stenosed pulmonary valve into the pulmonary artery, occasionally one can see a jet of blood as it impinges against the opposite wall of the pulmonary artery Furthermore, it is possible to measure the pressure in the right ventricle and in the pulmonary artery both before and after valvulotomy Measurement of the pressure after operation is of aid in order to make sure that the valve orifice is sufficiently enlarged so that the right ventricular pressure is reduced to normal or nearly to normal Nevertheless, if the pulmonary stenosis has been adequately relieved, failure of the right ventricular pressure to return to normal at operation is usually

not an indication for resection of the muscle at the base of the right ventricle In deed Engle et al [20] have shown that, after the relief of pulmonary stenosis, there may be a gradual drop in right ventricular pressure and over a period of months there may be a return to normal

In this operation the prime consideration is the relief of the pulmonary stenosis Even if prior to operation the patient has shown persistent cyanosis, when the pressure in the right auricle becomes normal, the valve covering the foramen ovale will close Therefore only if there is a gross defect in the auricular septum is closure of the defect necessary Unless the heart is greatly enlarged, the risk of operation is remarkably slight In 1953 the mortality rate, based on over 2,000 operations done in widely different places, was only 4 per cent

The results of operation can be judged by the change in the size of the heart in simple anterior posterior teleoroentgenograms and by the changes seen in the electrocardiogram Therefore cardiac catheterization is not necessary in order to determine the right ventricular pressure The more completely the pressure in the right ventricle is relieved, the more promptly the cardiac enlargement comes to an end [16] Except when the heart is greatly enlarged, if the right ventricular pressure drops to normal, the heart returns to normal size Landtman[17] has shown that the electrocardiogram also offers evidence of the relief of right ventricular strain in that the axis becomes balanced and over a period of months right ventricular dominance disappears Significant changes in the electrocardiogram are usually not apparent until six months after operation and it may be a year or longer before the maximum change is attained (see Figure XVII-19)

When the heart is enormously enlarged, the risk of operation is great and it is doubtful that perfection can be attained Nevertheless, the benefit may be tremendous The patient usually regains and maintains compensation, dyspnea disappears and his exercise tolerance greatly increases With lesser degrees of enlargement the heart will return to normal and the patient will be able to lead an entirely normal life

It is however, important to remember that the heart itself is not entirely normal The fibrotic valve is only cut it is not a normal valve Resection of the infundibular area does not restore the heart to normal The patient will remain susceptible to subacute bacterial endocarditis, therefore throughout his life he should receive the usual prophylactic treatment

PROGNOSIS

The prognosis varies with the severity of the pulmonary stenosis Mild degrees of pulmonary stenosis are compatible with a long and active life

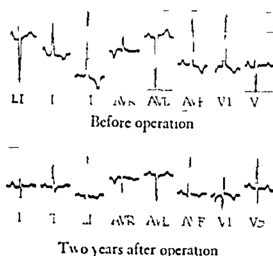


FIGURE XVII-19. Valvular pulmonary stenosis with an intact ventricular septum

Even moderate degrees of pulmonary stenosis may be compatible with longevity. Currens et al.[3] have reported the case of a man with severe pulmonary stenosis and patency of the foramen ovale who lived to be seventy years of age. The author has seen a specimen of a pulmonary stenosis from a man who served in the regular Army, contracted rheumatic fever late in life, and died of rheumatic heart disease when nearly seventy years of age. In this instance, in addition to mitral and aortic stenosis, autopsy revealed a cap-like stenosis of the pulmonary valve which reduced the lumen to approximately one third of its normal diameter. This degree of pulmonary stenosis had apparently never caused any difficulty.

If the pulmonary stenosis is sufficiently severe to place a strain on the right side of the heart, the strain can be relieved by operation. Since the development of cardiac surgery, most patients with this malformation may expect to enjoy long and active life.

Severe pulmonary stenosis greatly increases the work of the right side of the heart and unless the condition is corrected by surgery it leads to progressive cardiac enlargement, engorgement and pulsation of the liver, and eventually to edema and ascites. Patency of the foramen ovale lessens the strain on the heart but increases the oxygen unsaturation of the arterial blood and leads to polycythemia. Even for individuals with severe pulmonary stenosis who have not sought medical attention until after the heart is markedly enlarged, the life span may be greatly lengthened by successful operation.

SUMMARY

Pulmonary stenosis with an intact ventricular septum is a relatively common malformation Usually the stenosis is of the valvular type but in rare instances there may be an infundibular stenosis The pulmonary stenosis usually increases with age and leads to progressive cardiac enlargement The obstruction to the pulmonary orifice increases the work of the right ventricle and hence increases the pressure within the right ventricle and later that in the right auricle As the pressure in the right auricle rises if the valve covering the foramen ovale is not completely sealed, it will be forced open and a right to-left shunt will be established through it

The clinical findings vary with the severity of the pulmonary stenosis and with the structure of the foramen ovale

The patient is of a sturdy build

Dyspnea is the outstanding complaint Nevertheless, attacks of paroxysmal dyspnea are virtually unknown in infancy

The presence or absence of cyanosis depends upon the structure of the auricular septum If, as happens with 75 per cent of these patients, the foramen ovale is not completely sealed, cyanosis develops insidiously and generally becomes apparent between two and seven years of age

Polycythemia develops as the oxygen unsaturation of the arterial blood increases

Clubbing of the extremities gradually develops after cyanosis becomes apparent

The neck vessels may be engorged and show conspicuous pulsations

Enlargement of the liver occurs as the heart increases in size Pulsations at the margin of the liver are common, especially if the auricular septum is intact

Edema and ascites are late manifestations

Loss of consciousness is also a late manifestation

The size of the heart varies with the severity of the pulmonary stenosis With slight to moderate pulmonary stenosis there is no cardiac enlargement

Cardiac enlargement precedes symptoms therefore a patient suspected of this malformation should be kept under observation until he has attained his full growth Serial x rays give the best evidence of progressive cardiac enlargement in infancy whereas the electrocardiogram is the most sensitive index in childhood

A harsh systolic murmur and a thrill over the pulmonary area and in the

suprasternal notch, combined with a weak or absent second sound, are charac teristic of this malformation

Infundibular stenosis is to be suspected if the murmur is characteristic but there is no thrill in the suprasternal notch, it is also to be suspected if the mur mur is maximal in the third and fourth left interspaces, and the second sound at the base is clearly audible

The x ray shows fullness of the pulmonary conus and may show conspicuous pulsations in this area but minimal pulsations in the hilar shadows With tre mendous cardiac enlargement in early infancy or in adults with infundibular stenosis, there may be no poststenotic dilatation and hence no fullness of the pulmonary conus

The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy which becomes pronounced as the strain on the right ventricle increases

The circulation time is usually prolonged

The oxygen saturation of the arterial blood varies with the amount of venous blood shunted through the foramen ovale to the systemic circulation

Cardiac catheterization reveals high pressure in the right ventricle and low pressure in the pulmonary artery

Angiocardiography shows prompt opacification of the pulmonary artery, in which the dye lingers for a long time The aorta is not visualized until after the left auricle and the left ventricle have been delineated

The diagnosis is based upon the finding of a harsh systolic murmur and a thrill, a weak or absent pulmonic second sound, and electrocardiographic evi dence of a right axis deviation and right ventricular hypertrophy

The malformation calls for differentiation from an auricular septal defect, from primary pulmonary hypertension, occasionally from the Eisenmenger complex, and in some instances from a complete transposition of the great ves sels, from a tetralogy of Fallot, and from defective development of the right ventricle combined with pulmonary stenosis

Medical treatment, especially digitalis, is of great benefit if an infant develops cardiac failure in the neonatal period Digitalis should also be given to any pa tient who develops cardiac failure before he can reach a hospital where cardiac surgery is performed Inasmuch as the condition can be relieved by surgery, limitation of exercise is seldom necessary

Mild degrees of pulmonary stenosis may be compatible with a long and active life, therefore in such instances operation is not indicated On the other hand,

severe degrees of pulmonary stenosis lead to progressive cardiac enlargement and cardiac failure with edema and ascites

Operation is indicated if there is persistent cyanosis, cardiac enlargement or evidence of right ventricular strain or excessively high pressure in the right ventricle

Several types of operation have been devised All the operations give excellent results Inasmuch as the valve is not restored to normal, the risk of subacute bacterial endocarditis remains, therefore whenever indicated the patient should receive prophylactic chemotherapy throughout his life

For a patient who shows evidence of cardiac strain, operation removes the strain on the heart and changes the prognosis *from bad to excellent*

B *Stenosis in Either or Both Branches of the Pulmonary Artery*

In 1938 Oppenheimer[18] reported a case of stenosis of both main branches of the pulmonary artery and reviewed the literature No clinical attention was paid to the condition until 1954-55 In 1954 Søndergaard[19] reported the condition as coarctation of the pulmonary artery The following year Arvidsson et al [9] reported the occurrence of multiple stenoses of the pulmonary artery which they diagnosed by selective angiocardiography In the same year an additional case was reported by Kjellberg et al [1] In 1957 eight more clinical cases were reported by Gyllensward et al As in most instances, once an anomaly has been recognized it is detected with increasing frequency Furthermore, it produces a distinctive clinical syndrome

NATURE OF THE MALFORMATION

A localized constriction of the pulmonary artery may occur almost anywhere along its branches Gyllensward et al have emphasized that there are two main types Type I consists of a short membranous stenosis about 1 cm distal to the pulmonary valve and an elongated mild stenosis of one of the branches of the pulmonary artery and Type II is an extensive abnormality with multiple, short constrictions which usually involve the pulmonary arteries in both lungs Whether or not it is usual to have calcification in the pulmonary artery, as reported by Oppenheimer is not known, as most of the recent reports have been based on clinical observations and lack pathological confirmation

Actually there may be a single constriction in one main branch, bilateral constrictions in both main branches, or multiple constrictions scattered throughout

the branches of the pulmonary arteries Such constrictions affect the heart and circulation in a manner closely similar to that which occurs in valvular pulmonary stenosis, the only difference is that the obstruction in the pulmonary circulation lies beyond, instead of at, the pulmonary valve When the constriction is limited to one lung, it causes little or no strain on the circulation If, as more commonly occurs, it is bilateral, it places a strain on the circulation which is functionally similar to that caused by isolated valvular pulmonary stenosis Indeed, such a constriction of the pulmonary artery may occur in conjunction with valvular pulmonary stenosis Further, the abnormality may occur in combination with malformations which cause persistent cyanosis, under such circumstances each will contribute its own component It is the analysis of the separate components which will permit accurate diagnosis Hence this section is concerned with stenosis of the branches of the pulmonary artery when it occurs as an isolated abnormality

COURSE OF THE CIRCULATION

The course of the circulation is unaltered by the malformation unless the foramen ovale is held open by the high pressure in the right auricle Thus the *course of the circulation is fundamentally the same as that of isolated* valvular pulmonary stenosis (see Diagrams XVII–1 and 2)

PHYSIOLOGY OF THE MALFORMATION

The *constriction of the pulmonary artery affects* the circulation in a manner similar to that of any constriction elsewhere The pressure proximal to the obstruction is higher than that distal to it The narrowed area in the pulmonary artery obstructs the flow of blood to the lungs The pressure in the pulmonary artery proximal to the constriction is increased, hence more work is required of the right ventricle to pump the blood to the lungs Consequently there is right ventricular hypertrophy If the stenosis is sufficiently extreme, the high pressure in the right ventricle is transmitted back into the right auricle As the pressure in the right auricle rises, if the valve covering the foramen ovale is not completely sealed, it will be forced open and a right to-left shunt established at the auricular level

CLINICAL FINDINGS

The symptoms produced by this anomaly vary with the number and severity of the constrictions When there are bilateral constrictions in the pulmonary ar

teries, the clinical findings are similar to those in isolated valvular pulmonary stenosis

Cyanosis is usually absent If, however, the foramen ovale is not completely sealed when the pressure in the right auricle exceeds that in the left auricle, the valve will be forced open and a right *to-left shunt established* Such was the situation in the case reported by Oppenheimer

Dyspnea on exertion may occur because the obstruction to the pulmonary blood flow prevents the *increase in the circulation* to the lungs which normally occurs with exercise

CARDIAC FINDINGS

The heart may be normal in size and shape and there may be no murmurs over the precordium Since the work of the right ventricle is increased, the condition may lead to progressive cardiac enlargement *The second sound* over the pulmonary area is loud and banging, similar to that which occurs in pulmonary hypertension

A continuous murmur is audible over the lungs It has the humming quality similar to that produced by an anastomosis between a systemic artery and one of the branches of the pulmonary artery The continuous murmur is *due to the* continuous flow of blood from the area of high pressure proximal to the constriction to the area of low pressure distal to it Hence it is better heard when there is but a single constriction in one or both of the main branches of the pulmonary artery than when there are multiple constrictions throughout the lungs Indeed under the latter circumstance, the continuous murmur may be absent because the pressure gradient is gradually reduced between each of the successive constrictions

X RAY AND FLUOROSCOPIC FINDINGS

The contour of the heart is similar to that of 'pure pulmonary stenosis The prominence of the pulmonary conus is, however, not caused by poststenotic dilatation but by dilatation of the pulmonary artery proximal to *the stenosis* The multiple constrictions in the branches of the pulmonary artery reduce the size of these vessels Hence the vascular markings are decreased and the lungs appear clear

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram shows evidence of the increased work required of the

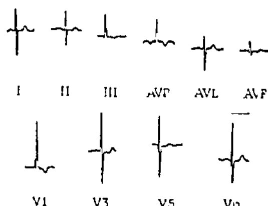


FIGURE XVII-20 Peripheral pulmonary stenosis (clinical diagnosis, not proven)

right ventricle The standard leads show a right axis deviation and the unipolar precordial leads show evidence of right ventricular hypertrophy (see Figure XVII-20)

SPECIAL TESTS

The red blood cell count, the amount of available hemoglobin, and the hematocrit reading are normal It is only in the rare instance when the foramen ovale is held open by the high pressure on the right side of the heart that the patient may develop polycythemia Usually, however, if polycythemia is present it is caused by some additional malformation

Cardiac catheterization shows an increased pressure in the right ventricle and in the pulmonary artery and no evidence of a shunt These findings are similar to those of primary pulmonary hypertension In order to demonstrate the constriction in the pulmonary artery the catheter must slip into the branch of the pulmonary artery in which the constriction lies and, furthermore, the constriction must not be so extreme that it is impossible to pass the catheter through it A number of investigators[20, 3, 24] have reported cases in which it was possible to demonstrate high pressure proximal to the constriction and an abrupt fall in the pressure when the catheter passed through the constriction

Angiocardiography may also reveal the condition[0, -] by the demonstration of abrupt narrowing in the branches of the pulmonary artery Proximal to the constriction the pulmonary artery and its branches are dilated and distended and there is an abrupt decrease in the size of the artery Arvidsson et al[0] have reported four cases diagnosed by selective angiocardiography

DIAGNOSIS

The diagnosis is based upon the clinical finding of a continuous murmur

over one or both lungs, combined with an accentuated pulmonic second sound It may be *confirmed* by cardiac catheterization or angiocardiography

DIFFERENTIAL DIAGNOSIS

The condition, when uncomplicated, may be confused with persistent patency *of the* ductus arteriosus, with primary pulmonary hypertension, *with* a hemi truncus arteriosus, or with other rare anomalies which cause a continuous murmur (see Chapter XXV)

Persistent patency of the ductus arteriosus causes a crescendo-decrescendo murmur of maximal intensity over the pulmonary area The second sound is audible within the continuous murmur In contrast to this, in the malformation under discussion, the pulmonic second sound is loud and banging and the continuous murmur is audible over the lungs

Primary pulmonary hypertension may be confused with this malformation because of the absence of murmurs over the precordium and the marked accentuation of the second sound over the pulmonary area, combined with evidence of high pressure in the right ventricle and in the pulmonary artery The presence of a continuous murmur over the lungs differentiates the two conditions

A hemi truncus arteriosus may readily be confused with localized constrictions in the pulmonary artery Both are rare anomalies which cause a continuous murmur over the lungs A hemi truncus arteriosus causes no strain on the right side of the heart (see Chapter XIV, Section B) Indeed, the right ventricle pumps the blood to only one lung whereas when the pulmonary artery is constricted, the work of the right ventricle is increased Hence the electrocardiogram aids in the differentiation of the two conditions

The malformation under discussion must also be differentiated from the other rare anomalies which cause a continuous murmur (see under Differential Diagnosis in Chapter XXV)

When there is persistent cyanosis this malformation calls for differentiation from a truncus arteriosus with relatively adequate pulmonary blood flow Cardiac catheterization or angiocardiography shows that the pulmonary artery, not the aorta or the truncus, arises from the right ventricle and there is no evidence of a shunt in the right ventricle

TREATMENT

As of 1960 there has been no satisfactory treatment Although in theory the *constricted* area could be resected as it is in coarctation of the aorta, the occur

rence of multiple constrictions renders such an operation difficult if not impossible

PROGNOSIS

The prognosis depends upon the severity of the condition. Although the author has seen one baby with severe bilateral constrictions in the main branches of the pulmonary artery who died of right sided cardiac failure in early infancy,[18] the condition is generally far less severe and is usually compatible with relative longevity

SUMMARY

Stenosis may occur in either or both branches of the pulmonary artery. There may be single or multiple stenoses. Severe constriction of the pulmonary artery increases the pressure against which the right ventricle must work. The condition may lead to progressive right sided cardiac enlargement. The second heart sound over the pulmonary area is accentuated and there may be a continuous murmur audible over the lungs. The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy. Cardiac catheterization shows high pressure in the right ventricle and in the main pulmonary artery. If it is possible to pass the catheter through the constriction, the pressure distal to the constriction will be lower than that proximal to it. It may be possible to demonstrate the area of constriction by angiocardiography

The condition requires differentiation from persistent patency of the ductus arteriosus, pulmonary hypertension, a hemi truncus arteriosus, and other causes of a continuous murmur. Multiple peripheral pulmonary stenoses are difficult to correct

The prognosis varies with the severity of the condition. When the stenosis is severe, the prognosis is guarded. Less severe stenoses are compatible with relative longevity

References

1 Brock, R C Pulmonary valvulotomy for the relief of congenital pulmonary stenosis report of 3 cases Brit M J 1 1121–1126 1948

2 Keith, A The Hunterian Lectures on malformations of the heart Lancet 2 359–363, 433–435 519–523 1909

3 Currens, J H, T D Kinney and P D White Pulmonary stenosis with intact interventricular septum report of 11 cases Am Heart J 30 491–510, 1945

4 Engle M A and H B Taussig Valvular pulmonic stenosis with intact ventricular septum and patent foramen ovale report of illustrative cases and analysis of clinical syndrome Circulation 2 481–493 1950
5 Potts W J S Smith and S Gibson Anastomosis of the aorta to a pulmonary artery certain types in congenital heart disease J.A.M.A 132 627–631 1946
6 Campbell M Simple pulmonary stenosis, pulmonary valvular stenosis with a closed ventricular septum Brit Heart J 16 273–300 1954
7 Engle M A T Ito, D S Lukas and H P Goldberg Electrocardiographic evaluation of pulmonic stenosis Society of Pediatric Research May 1959
8 Bing R J W Reber J E Sparks F A Balboni, A G Vitale and M Hanlon Congenital pulmonary stenosis J.A.M.A 154 127–129 1954
9 Beard, E F D A Cooley and J R Latson Combined congenital subaortic stenosis and infundibular subpulmonic stenosis report of a case with successful surgical treatment A.M.A Arch Int Med 100 647–650 1957
10 Swan H Stenosis of the pulmonic and aortic valves postgraduate course in cardiovascular surgery 43rd Annual Clinical Congress of the American College of Surgeons October 1957
11 Sissman N J C A Neill F C Spencer and H B Taussig Congenital aortic stenosis Circulation 19 458–468 1959
12 Potts W J., S Gibson W L Riker and C R Leininger Congenital pulmonary stenosis with intact ventricular septum J.A.M.A 144 8–12 1950
13 Blount S G Jr M C McCord H Mueller and H Swan Isolated valvular pulmonic stenosis clinical and physiologic response to open valvuloplasty Circulation 10 161–172 1954
14 Søndergaard T Valvulotomy for pulmonary stenosis performed through the main stem of the pulmonary artery utilizing a special ring clamp Acta chir scandinav 104 362–372 1953
15 Engle M A C R Holswade H P Goldberg D S Lukas and F Glenn Regression after open valvotomy of infundibular stenosis accompanying severe valvular pulmonic stenosis Circulation 17 862–873 1958
16 Hosier D M J L Pitts and H B Taussig Results of valvulotomy for valvular pulmonary stenosis with intact ventricular septum analysis of 69 patients Circulation 14 9–16 1956
17 Landtman B Postoperative changes in the electrocardiogram in congenital heart disease pure pulmonary stenosis Circulation 10 839–870 1954
18 Oppenheimer E H Partial atresia of the main branches of the pulmonary artery occurring in infancy and accompanied by calcification of the pulmonary artery and aorta. Bull Johns Hopkins Hosp 63 261–277 1938
19 Søndergaard T Coarctation of the pulmonary artery Danish M Bull 1 46–48 1954
20 Arvidsson H, J Karnell and T Moller Multiple stenosis of the pulmonary arteries associated with pulmonary hypertension diagnosed by selective angiocardiography Acta radiol 44 209–216 1955

21 Kjellberg S R, E Mannheimer, V Rudhe, and B Jonsson Diagnosis of Congenital Heart Disease Chicago, Year Book Publishers, 1955

22 Gyllenswärd, A, H Lodin, A Lundberg, and T Moller Congenital multiple peripheral stenoses of the pulmonary artery Pediatrics 19 399–410, 1957

23 Coles, J E and W J Walker Coarctation of the pulmonary artery Am Heart J 52 469–473, 1956

24 Williams, C B, R L Lange and H H Hecht Postvalvular stenosis of the pulmonary artery Circulation 16 195–199, 1957

CHAPTER XVIII

PULMONARY HYPERTENSION

PULMONARY hypertension may be 'primary' or it may be 'secondary.' Primary pulmonary hypertension means that the initial pathology lies in the pulmonary vascular bed and that the heart is normal except for the strain placed on it by the increased peripheral resistance in the pulmonary vascular bed. In contrast to this, 'secondary' pulmonary hypertension means that the high pressure in the pulmonary artery is secondary to a malformation of the heart. Edwards[1] has pointed out that in a number of malformations of the heart the blood is ejected to the lungs under systemic pressure. Under such circumstances the high pressure in the lungs is secondary to the cardiac malformation. A brief review of the normal changes in the lungs which occur immediately after birth will help to clarify the nature of this pathology.

Civin and Edwards made a detailed study of microscopic sections of the lungs of patients from birth to old age. These authors followed Brenner's classifications of the division of the intrapulmonary arteries into three main groups: (1) elastic arteries, (2) muscular arteries, and (3) arterioles. The investigators demonstrated that in the fetus the lumen of muscular arteries of the lungs is narrow and the media is thick, the adventitia is thick and the intima thin. Normally during the first six months of extra-uterine life the lumen of the muscular arteries widens and the media becomes thin. Thus the lumen of the arteries increases in size and the thickness of the muscular wall decreases. The adventitia becomes much thinner and the intima remains unchanged. Such are the normal changes which occur during the first months of life. Moreover, the pulmonary vascular bed continues to expand up to about the twentieth year of life. The greatest changes, however, normally occur in early infancy.

Edwards has postulated that, since no well-established vasomotor response in the pulmonary vascular bed has been demonstrated, the narrow lumina of the muscular arteries constitute one factor which increases the resistance to the pulmonary flow during fetal life, and, furthermore, that the gradual increase in the size of these lumina lowers the pulmonary resistance. He has found that the principal changes in the pulmonary vascular bed which occur in the first few months of extra-uterine life are the widening of the lumina of the muscular arteries and the thinning of the media of these vessels.

Furthermore, Edwards has pointed out that in a number of malformations, nainly those in which the blood is pumped into the pulmonary artery under ystemic pressure, the pressure in the pulmonary artery is a vital factor in the listribution of the blood to the two circulations Were the pulmonary pressure ow, the pulmonary blood flow would be so excessive that the patient would lrown in his lungs Edwards has reported narrowing of the intrapulmonary essels in a patient with an Eisenmenger complex[1, 3] and also in two patients in vhom the ductus arteriosus opened into the descending aorta distal to the co- rctation[4] The muscular arteries were characterized by a thick media and a rela ively narrow lumen In older patients he has observed that, in addition to the hick media, intimal fibrosis develops He believes that, in malformations of the ieart in which there is excessive pulmonary blood flow, the muscular arteries naintain their fetal characteristics of a small lumen and a thick media as a com *ensatory mechanism These changes initially are caused by a delay in the open* ng up of the pulmonary vascular bed which protects the lungs from an excessive ulmonary blood flow under abnormally high pressure Edwards conceives that he initial changes are reversible in that, if the excessive pressure were removed, he lungs would be capable of normal expansion The long-continued high pres- ure ultimately leads to intimal proliferation and fibrosis These changes rigidly ix and further constrict the lumina of the vessels and thus further increase the esistance to the pulmonary blood flow When such changes occur, the condition s no longer reversible Thus, although in the early stages pulmonary hyperten ion is compensatory and the changes are reversible, in the later stages changes ccur which are progressive and irreversible

Edwards has presented strong evidence to show that such is the sequence of vents in malformations of the heart in which blood is ejected simultaneously nto the systemic circulation and the pulmonary circulation Pulmonary hyper ension always occurs in an Eisenmenger complex, in a single ventricle without ulmonary stenosis, in a truncus arteriosus in which large pulmonary arteries rise from the base of the truncus, and in a Taussig Bing heart Whenever the ight ventricle pumps the blood to the systemic circulation through the ductus rteriosus, the pressure in the pulmonary artery must be the same as that in the ystemic circulation, such is the situation in aortic atresia or marked hypoplasia f the *ascending aorta when the systemic circulation is* supplied from the pul nonary artery through the ductus arteriosus

In addition to these conditions in which pulmonary hypertension is in a sense n integral part of the malformation of the heart, or at least is the inevitable re

sult of the cardiac abnormality, there is a condition in which, in some instances, the pulmonary hypertension appears to be primary, as for example in *cor pulmonale* in which the changes in the heart appear to be secondary to the high pressure in the lungs Under such circumstances the heart is essentially normal Nevertheless, the increased resistance in the lungs affects the heart and circulation quite as profoundly as does a coarctation of the aorta The principal difference is that the pulmonary hypertension places a strain on the right side of the heart, whereas coarctation of the aorta places a strain on the left side

ETIOLOGY

The etiology is obscure Indeed, the author believes that there may be a number of different causes of primary pulmonary hypertension In some instances the increased pulmonary pressure appears to be secondary to some pulmonary infection or to the inhalation of some irritating substance In other instances multiple thrombi have been found in the smaller pulmonary vessels and occasionally there is a massive thrombus in the main branches of the pulmonary artery In still other instances it appears to be secondary to long standing pulmonary disease and asthma Pulmonary hypertension is also seen in severe kyphoscoliosis It is possible that there may be a pulmonary arteriolar disease which is comparable to systemic arteriolar sclerosis Cor pulmonale may appear as an acute disease or as a chronic illness The disease is notoriously common in and around Pittsburgh, where it seems as if it were an occupational disease It is also known to occur in families, as for example in the family referred to the author by Dr Dresdale,[5] where the mother, her sister, and her son all died of pulmonary hypertension In the family reported by Coleman et al[6] three siblings suffered from primary pulmonary hypertension Furthermore, the author has studied patients of all ages, from six months to over twenty years, with what appeared to be primary pulmonary hypertension

The pathology in the lungs is not the same in all instances There is one group of cases in which the minute vessels in the capillaries of the lungs show an endarteritis and inflammatory changes In another group of cases the lesions appear to lie in the small muscular arteries and numerous thrombi occur in the lungs, many of which show evidence of recanalization These changes are similar to those described by Rich[7] in patients with long standing polycythemia but have also been observed in a number of infants in whom there was no polycythemia Such lesions are being reported with increasing frequency in adults with cor pulmonale

It is also conceivable that 'primary' pulmonary hypertension may be due to an arrest in the development of the pulmonary vascular bed of such a nature that the intrapulmonary arteries fail to undergo their normal postnatal involution It may be that, in the presence of a congenital abnormality of the pulmonary vascular bed, there are two opposing factors one is an abnormality of the lungs which is of such a nature as to increase the peripheral resistance and the other is the gradual expansion of the pulmonary vascular bed which, as Edwards has shown, normally continues up to twenty years of age Thus, during childhood and adolescence, the latter may offset the former with the result that the increased pulmonary resistance may not become manifest until early adult life Furthermore, it is possible that in some instances an acquired pulmonary infection which would produce insignificant scarring in a normal adult may be sufficient to swing the balance unfavorably in a patient with an abnormality in the pulmonary vascular bed

It is notoriously true, and has been repeatedly emphasized, that although patients with cor pulmonale give a history of repeated respiratory infections, the condition is rarely seen in chest clinics among the patients who are known to suffer from long standing or chronic pulmonary infections

Allergy appears to be a factor in some instances, as in one of the author's patients who was extremely short of breath, so short of breath that she panted over the telephone After a few weeks in Maine this patient was able to enjoy swimming and, when she returned, the dyspnea was no longer detectable over the telephone

Although it has long been assumed that there is no nervous control of the lungs, there is increasing evidence that such a mechanism may exist Lilienthal and Riley,[8] in their review of diseases of the respiratory system, referred to the possibility and discussed the evidence in favor of it Indeed, the response of older patients with an Eisenmenger complex to the inhalation of oxygen also strongly suggests the existence of some nervous or humoral control over the pulmonary vascular bed

As previously mentioned, the author has studied a number of infants who suffered from primary" pulmonary hypertension When the difficulty dates from birth, it seems clear that the pulmonary hypertension is congenital in origin Thus it appears that pulmonary hypertension may be congenital or may result from acquired disease

Regardless of etiology, 'primary' pulmonary hypertension occurs as an iso-

lated abnormality and may be present from birth The increased resistance in the pulmonary vascular bed increases the work required of the right ventricle and this in turn increases the work required of the right auricle As in all conditions which place a strain on the right side of the heart, the foramen ovale may be held open by the high pressure in the right auricle In contrast to the abnormality in valvular pulmonary stenosis with an intact ventricular septum, and in Ebstein s anomaly of the tricuspid valve, the difficulty lies not within the heart but in the lungs, consequently the high pressure in the pulmonary artery may tend to keep the ductus arteriosus from closing Thus primary pulmonary hypertension occurs as an isolated anomaly and it also occurs with persistent patency of the ductus arteriosus, the latter combination of anomalies produces a distinctive clinical syndrome and is discussed in Section B Section A is concerned with primary pulmonary hypertension with and without patency of the foramen ovale

A Primary Pulmonary Hypertension with and without Patency of the Foramen Ovale

NATURE OF THE MALFORMATION

This malformation is primarily due to changes in the pulmonary vascular bed The peripheral pulmonary vascular bed is abnormally constricted and thereby the peripheral resistance in the lungs is increased and the intrapulmonary arterial pressure is abnormally high The heart itself is normally formed The ductus arteriosus is closed Although the nature of the changes in the lungs is not fully understood, and indeed may not always be the same, any condition which increases the peripheral resistance in the pulmonary vascular bed increases the work of the right ventricle The right ventricle becomes slightly dilated and greatly hypertrophied The strain on the right ventricle may be so great that the pulmonary valve becomes insufficient. The increased pressure in the right ventricle increases the work required of the right auricle to pump the blood into the right ventricle consequently the right auricle is also hypertrophied Inasmuch as the heart is normal, if the foramen ovale has not been completely sealed, the valve will be forced open by the high pressure in the right auricle and thereby a right to-left shunt will be established This occurs in approximately 75 per cent of all patients in whom the pressure in the right auricle is abnormally high during early infancy

COURSE OF THE CIRCULATION

If the foramen ovale becomes completely sealed and the ductus arteriosus undergoes normal obliteration, the course of the circulation is normal The blood flows from the right auricle to the right ventricle and thence is pumped out through the pulmonary artery to the lungs The constriction of the pulmonary vascular bed increases the peripheral resistance in the lungs and this in turn increases the work of the right side of the heart Nevertheless, the blood which passes through the lungs is fully oxygenated and is returned in the normal fashion to the left auricle, thence it flows into the left ventricle and is pumped out through the aorta to the systemic circulation It follows that the blood in the systemic circulation is fully oxygenated The blood is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle, there the cycle starts again (see Diagram XVIII-1)

As with all malformations which place a strain on the right side of the heart, as the pressure in the right ventricle rises, the right auricle cannot empty itself as readily as normally Consequently, if the foramen ovale is not completely sealed, as the pressure within the right auricle rises, the increased pressure in that chamber eventually will force the valve covering the foramen ovale to open and some venous blood from the right auricle is shunted into the left auricle Under such circumstances the mixture of oxygenated blood which is returned to the left auricle from the lungs and of venous blood which is shunted from the right auricle to the left auricle, will flow into the left ventricle and be pumped out through the aorta into the systemic circulation The oxygen saturation of the arterial blood will be reduced directly in proportion to the volume of venous blood which is shunted from the right auricle through the foramen ovale to the systemic circulation When the volume of unoxygenated blood so shunted becomes sufficiently great, the patient will show cyanosis (see Diagram XVIII-2)

PHYSIOLOGY OF THE MALFORMATION

The physiology of the malformation concerns the abnormality in the lungs There is progressive narrowing of the small arteries and arterioles in the pulmonary vascular bed The pulmonary capillary bed usually remains normal and hence the pressure in the capillaries remains normal Nevertheless, the increased resistance in the pulmonary vascular bed increases the work required of the right ventricle and consequently increases the pressure in that chamber The increased pressure in the right ventricle increases the pressure against which the right auricle must work and this in turn raises the pressure in that chamber If the fora

men ovale is not completely sealed, it acts as an escape valve and slightly reduces the strain on the heart If it is entirely sealed, the escape valve is closed and cardiac failure occurs early The peripheral resistance is frequently so greatly increased that the pressure in the pulmonary circulation exceeds the systemic pressure Unless the pressure can be relieved the condition leads inevitably to progressive right sided cardiac enlargement and cardiac failure

CLINICAL FINDINGS

The clinical findings vary with the severity of the condition In some instances the condition may be so severe that the infant dies of cardiac failure before one year of age, in others the condition may be compatible with life for more than thirty years

Difficulty in feeding and *failure to gain* are common complaints in an infant with severe pulmonary hypertension

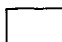
Repeated respiratory infections bronchitis and *pneumonia* frequently occur during infancy and childhood The patient may, however, be asymptomatic in infancy and childhood and no abnormality may be suspected until early adult life, when he suffers from some pulmonary infection and thereafter develops dyspnea

Dyspnea is the outstanding complaint In infancy rapid respiration is the initial manifestation Not infrequently the condition progresses to cardiac failure before the seriousness of the respiratory distress is appreciated In an older patient the onset of dyspnea may occur abruptly after an intercurrent pulmonary infection and become rapidly worse until he is so short of breath that conversation causes him to pant

Attacks of suffocation occasionally occur The pulmonary artery may be so large and the pressure in it so high that either of its main branches may compress the bronchi The author has seen one child in whom the compression was so severe that he suffered from severe dyspnea after lying for a short time flat in bed Bronchoscopic examination revealed compression of the right or left main bronchus, depending upon which side the child was lying

The presence or absence of cyanosis depends upon the structure of the foramen ovale Infants with severe pulmonary hypertension almost always show persistent cyanosis and oxygen unsaturation of the arterial blood, because the high pressure in the right auricle tends to keep the foramen ovale open and a right-to-left shunt is thereby established If the foramen ovale is completely sealed, the patient will show no cyanosis The oxygen saturation of the arterial blood is

Diagram XVIII-1



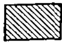
Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

DIAGRAM XVIII–1

Primary pulmonary hypertension

In this malformation the primary abnormality is the increased resistance in the pulmonary vascular bed the ductus arteriosus is obliterated and the foramen ovale is sealed The circulation of the blood is normal

The blood from the right auricle flows into the right ventricle and is pumped out through the pulmonary artery to the lungs, where it is oxygenated The blood is returned in the normal manner to the left auricle thence it flows to the left ventricle and is pumped out to the systemic circulation and returned by the superior vena cava and the inferior vena cava to the right auricle There the cycle starts again

The only cardiac change which occurs is the hypertrophy of the right ventricle and the right auricle which is secondary to the increased work required of the right side of the heart to pump the blood to the lungs against the increased resistance in the pulmonary vascular bed

Clinical diagnosis Dyspnea is the outstanding complaint The heart is usually not greatly enlarged The pulmonic second sound is markedly accentuated There may be a loud early diastolic murmur The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

Diagram xviii–2

Head and upper extremities

R Lung

L Lung

P.A.

P.A.

S.V.C.

P.V.

P.V.

Aorta

Pulmonary Artery

L.A.

R.A.

L.V.

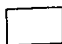
R.V.

Foramen Ovale

Descending Aorta

I.V.C.

Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

DIAGRAM XVIII–2

Primary pulmonary hypertension with patency of the foramen ovale

In this malformation primary pulmonary hypertension is combined with patency of the foramen ovale. The foramen ovale acts as an escape valve for the high pressure in the right auricle by the establishment of a right to left shunt

Under such circumstances although most of the blood in the right auricle flows into the right ventricle and is pumped out through the pulmonary artery to the lungs and is returned in the normal fashion to the left auricle some blood from the right auricle is shunted through the foramen ovale to the left auricle Consequently the left auricle receives fully oxygenated blood from the lungs and some venous blood from the right auricle This mixture of oxygenated and venous blood flows into the left ventricle and is pumped out through the aorta to the systemic circulation and is returned by the superior and inferior venae cavae to the right auricle There the cycle starts again

Clinical diagnosis Cyanosis usually develops insidiously It depends on the volume of venous blood shunted into the systemic circulation, hence cyanosis may be apparent at birth or may not develop until adolescence or adult life The heart is of approximately normal size with an insignificant systolic murmur and a markedly accentuated second sound over the pulmonary area The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

pproximately normal In an older patient who shows arterial oxygen unsatura on it may seem plausible that the unsaturation could be caused by the pathology the lungs, actually in almost every instance in which a patient has shown anosis, autopsy has revealed patency of the foramen ovale Therefore it seems obable that the cyanosis in primary pulmonary hypertension has the same igin as that in "pure" pulmonary stenosis and in Ebstein's anomaly of the tri spid valve namely, the cyanosis is caused by the shunting of venous blood om the right auricle through the foramen ovale to the systemic circulation

Clubbing is less marked than is the cyanosis

Polycythemia develops gradually as the patient suffers from persistent oxygen saturation of the arterial blood

Hemoptysis is a common complaint in older patients but seldom if ever oc rs in infancy

The blood pressure is normal The *pulse* is of equal strength in the arm and e leg

Engorgement of the liver occurs as the heart begins to fail The liver may be me enormously enlarged and frequently pulsations are palpable at its margin ngorgement of the liver occurs readily in patients who show no cyanosis, be use the foramen ovale is completely or nearly completely sealed

Edema of the extremities is also a late manifestation Inasmuch as the condi on is progressive, the edema may be extremely troublesome and difficult to treat

Ascites is also a late manifestation and may become extreme

Syncope is a late and serious manifestation The circulation through the ngs is so slow that the minute output of the heart is reduced Consequently ere is not only a low pulmonary blood flow but also a low systemic flow Fi lly, the circulation of the blood becomes so sluggish that the supply of oxygen the brain is seriously impaired Such appears to be the cause of the attacks of ncope from which these patients suffer After the development of syncopal at cks the duration of life is relatively short Many a patient dies during an attack syncope

Precordial pain may occur but is rare It, too, is a late manifestation and is nerally considered to be due to coronary insufficiency, it goes hand in hand th myocardial failure

RDIAC FINDINGS

The size of the heart varies with the severity of the condition and the age at uch it develops When severe pulmonary hypertension dates from birth, the

heart undergoes progressive enlargement and cardiac failure occurs at an early age When the pulmonary hypertension is less severe or develops at a later age, the heart may be but slightly enlarged Over a period of years the heart undergoes gradual progressive enlargement The pulmonary vascular disease may develop so rapidly that the condition is fatal before the heart becomes enormously enlarged

The second sound at the base of the heart to the left of the sternum is markedly accentuated and often reduplicated A decrease in the intensity of the second sound may, however, occur in the presence of cardiac failure, it is indicative of myocardial weakness

A systolic murmur may or may not be present Usually it is not loud

Pulmonary insufficiency frequently develops in children and young adults Indeed, the finding of a loud early diastolic murmur along the left sternal border in a patient with a markedly accentuated pulmonic second sound should always arouse suspicion of pulmonary hypertension The intensity of the diastolic murmur varies greatly with the patient s state of compensation If the condition is not too severe, a brief period of bed rest may cause the murmur to disappear, as the disease progresses, the patient may develop a systolic as well as a diastolic murmur The author has studied one seven year-old child in whom the murmur closely resembled the harsh continuous murmur of a patent ductus but was maximal in the third left interspace After two days of digitalis and rest in bed the continuous murmur disappeared and was replaced by an inconstant diastolic murmur There are few, if any, other cardiac abnormalities which show such variability of murmurs over such a short period of time

X RAY AND FLUOROSCOPIC FINDINGS

The heart may or may not be enlarged The pulmonary conus is full and usually the main pulmonary arteries are markedly enlarged Figure XVIII–1 shows great cardiac enlargement in a child of nine years Figure XVIII–2 shows the contour of the heart of a boy of thirteen with severe primary' pulmonary hypertension Fluoroscopy usually reveals a slight hilar dance and the peripheral lung fields are clear Examination in the oblique positions gives little additional information There is no enlargement of the left auricle, hence the esophagram is normal

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a marked right axis deviation and the unipolar pre



Figure xviii-1 Primary pulmonary hypertension Child

cordial leads show evidence of right ventricular hypertrophy or the pattern of so-called right ventricular "strain" (see Figure xviii-3)

SPECIAL TESTS

The circulation time is normal or prolonged

Cardiac catheterization reveals high pressure on the right side of the heart The pressure in the right ventricle is elevated and the pressure in the pulmonary artery is as high or higher than that in the right ventricle but the pulmonary capillary pressure is usually normal If the foramen ovale is closed, there is no shunt Under such circumstances the oxygen content of the blood samples taken from the right auricle, the right ventricle, and the pulmonary artery should be identical Even if the foramen ovale is patent, the shunt is from right to left, there should be no significant difference in the oxygen content of the blood in the various places in the right side of the heart Cardiac catheterization is, however, not without risk, as these patients are extremely susceptible to arrhythmias



FIGURE XVIII-2 Primary pulmonary hypertension Child

Nevertheless, the procedure is more informative and less dangerous than angiocardiography

Angiocardiography is not helpful and may be dangerous The constriction in the pulmonary arteriolar bed slows the circulation of the blood through the lungs If the pulmonary hypertension is marked, the blockage of the pulmonary arterioles with the dye may entirely cut off the supply of oxygen to the patient and lead to death The only positive finding upon angiocardiography is the lingering of the contrast substance in the right ventricle and the main pulmonary artery Thus the picture is identical with that of valvular pulmonary stenosis with an intact ventricular septum

DIAGNOSIS

The diagnosis is based on the marked accentuation of the second sound at the base of the heart to the left of the sternum combined with evidence of right ventricular hypertrophy and strain The occurrence of a loud early diastolic murmur is common The x ray shows fullness of the pulmonary conus and the

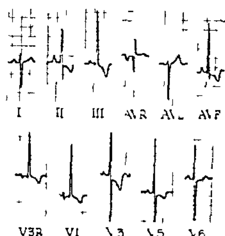


FIGURE XVIII-5 Primary pulmonary hypertension Child

main pulmonary arteries, upon fluoroscopy a hilar dance is visible Cardiac catheterization reveals a high pressure in the main pulmonary artery and no evidence of an intracardiac shunt or abnormality

DIFFERENTIAL DIAGNOSIS

In young infants who show cyanosis, the condition may simulate a tetralogy of Fallot, a complete transposition of the great vessels, or "pure" pulmonary stenosis, that is, valvular pulmonary stenosis in which the ventricular septum is intact and the foramen ovale is held open by the high pressure on the right side of the heart

In children and young adults, prior to the development of cyanosis, the condition may be confused with idiopathic dilatation of the pulmonary artery, a corrected transposition of the great vessels, an auricular septal defect, a patent ductus arteriosus without a continuous murmur, or acquired heart disease with *rheumatic or syphilitic aortic insufficiency, and occasionally with aortic stenosis* When the onset of cyanosis is delayed, the condition requires differentiation from polycythemia vera and the Eisenmenger complex With or without cyanosis the malformation must be differentiated from 'pure' pulmonary stenosis or Ebstein s anomaly of the tricuspid valve The condition may occasionally be confused with mitral stenosis and with a tumor of the left auricle

Tetralogy of Fallot may be confused with primary pulmonary hypertension in young infants who show deep persistent cyanosis and no murmurs Over a period of months, the evidence of progressive cardiac enlargement clearly differentiates the malformation under discussion from a tetralogy of Fallot

A complete transposition of the great vessels may also be confused with pri

nary pulmonary hypertension, as at birth in both instances the heart is normal in size and the vascular markings are slightly increased If there is pulmonary hypertension, as the heart undergoes enlargement the vascular markings decrease and the fullness of the pulmonary conus becomes apparent, thereby differentiating the condition from a complete transposition of the great vessels

Pure pulmonary stenosis resembles primary pulmonary hypertension in that progressive right sided cardiac enlargement and fullness of the pulmonary conus may occur, and there may or may not be cyanosis In pure pulmonary stenosis the systolic murmur is loud and harsh and ends abruptly with the end of systole and the second sound is weak or absent, whereas in primary pulmonary hypertension the second sound over the pulmonary area is markedly accentuated Indeed, it is only in the presence of cardiac failure and the consequent weakening of the second sound that the two conditions may be confused Under such circumstances cardiac catheterization may be necessary to establish the diagnosis

Idiopathic dilatation of the pulmonary artery differs from primary pulmonary hypertension in that the pulmonic second sound is not accentuated The electrocardiogram shows no evidence of extreme right ventricular hypertrophy Cardiac catheterization confirms the clinical impression that the pulmonary pressure is normal

Corrected transposition of the great vessels may be confused with 'primary' pulmonary hypertension because of the finding of marked accentuation of the second heart sound over the pulmonary area combined with x ray evidence of fullness of the pulmonary conus In a corrected transposition of the great vessels the electrocardiogram shows a tendency to a left axis deviation and evidence of left ventricular hypertrophy Furthermore, angiocardiography clearly shows that the fullness of the pulmonary conus is caused by the ascending aorta

An auricular septal defect is usually associated with a systolic murmur over the precordium which is frequently well heard posteriorly The pulmonic second sound is reduplicated but it is not as markedly accentuated as in patients with primary pulmonary hypertension The electrocardiogram shows a tendency to a right axis deviation, and commonly shows evidence of a right bundle branch block, rather than the pattern of extreme right ventricular hypertrophy

A patent ductus arteriosus may be mistaken for primary pulmonary hypertension especially in young infants while the pulmonary pressure remains high, that is before the development of a continuous murmur The heart is usually markedly enlarged, a systolic murmur and often a low pitched mid diastolic

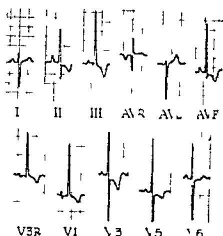


FIGURE XVIII-3 Primary pulmonary hypertension Child

main pulmonary arteries, upon fluoroscopy a hilar dance is visible Cardiac catheterization reveals a high pressure in the main pulmonary artery and no evidence of an intracardiac shunt or abnormality

DIFFERENTIAL DIAGNOSIS

In young infants who show cyanosis, the condition may simulate a tetralogy of Fallot, a complete transposition of the great vessels, or "pure" pulmonary stenosis, that is, valvular pulmonary stenosis in which the ventricular septum is intact and the foramen ovale is held open by the high pressure on the right side of the heart

In children and young adults, prior to the development of cyanosis, the condition may be confused with idiopathic dilatation of the pulmonary artery, a corrected transposition of the great vessels, an auricular septal defect, a patent ductus arteriosus without a continuous murmur, or acquired heart disease with rheumatic or syphilitic aortic insufficiency, and occasionally with aortic stenosis When the onset of cyanosis is delayed, the condition requires differentiation from polycythemia vera and the Eisenmenger complex With or without cyanosis the malformation must be differentiated from 'pure' pulmonary stenosis or Ebstein's anomaly of the tricuspid valve The condition may occasionally be confused with mitral stenosis and with a tumor of the left auricle

Tetralogy of Fallot may be confused with primary pulmonary hypertension in young infants who show deep persistent cyanosis and no murmurs Over a period of months, the evidence of progressive cardiac enlargement clearly differentiates the malformation under discussion from a tetralogy of Fallot

A complete transposition of the great vessels may also be confused with pri

sounds are weaker and more confused The electrocardiogram does not show a marked right axis deviation and seldom shows evidence of right ventricular hypertrophy in V_1

Mitral stenosis is occasionally so severe that too little blood is forced through the mitral valve to cause a presystolic murmur Only a faint systolic murmur is audible over the base of the heart and the pulmonary second sound is accentuated X ray or fluoroscopic examination should reveal enlargement of the left auricle The electrocardiogram usually shows notched P waves associated with left auricular enlargement, and not the "pyramidal" P waves seen with dilatation and hypertrophy of the right auricle Cardiac catheterization not only shows high pressure in the right ventricle and in the pulmonary artery but also reveals a high wedge pressure

A tumor of the left auricle is rare Nevertheless, since the clinical syndrome[8] produced thereby resembles mitral stenosis, it too may require differentiation from primary pulmonary hypertension A tumor of the left auricle which obstructs the flow of blood through the mitral valve may give the clinical and laboratory signs of mitral obstruction without circulatory signs of mitral stenosis Fatigue is a common complaint There may be dyspnea on exertion and acute attacks of pulmonary edema Furthermore, the obstruction to the flow of blood through the left auricle causes back pressure in the lesser circulation, this eventually increases the pressure in the pulmonary artery and causes right sided cardiac strain Cardiac catheterization shows not only high pressure in the pulmonary artery but increased pressure in the pulmonary capillary bed Angiocardiography may be of aid in that it may demonstrate a filling defect in the left auricle

COMPLICATIONS

The complications result from the low pulmonary blood flow with the correspondingly low systemic flow and from the polycythemia As the circulation to the lungs becomes extremely sluggish, thrombosis may develop in the main branches of the pulmonary artery Such thrombi may become so enormous that they seriously obstruct the pulmonary blood flow and thereby still further increase the work required of the right ventricle Thrombosis of one of the main branches of the pulmonary artery should be suspected when there is enormous dilatation of the hilar vessels and minimal or no pulsation visible therein

Terminally the systemic circulation may become so sluggish that multiple systemic thrombi develop

murmur are present The lungs appear extremely vascular There is usually either a balanced electrocardiogram or a tendency to a left axis deviation with evidence of "combined" ventricular hypertrophy

Rheumatic or syphilitic heart disease and aortic insufficiency are considered only because of the occurrence of a diastolic murmur along the left sternal border The absence of any history of acute rheumatic fever or of syphilitic infection, combined with electrocardiographic evidence of right rather than left ventricular hypertrophy, should offer the clue to the diagnosis Furthermore, fluoroscopic examination will reveal *no fullness of the pulmonary conus* and slight, if any, hilar pulsations In doubtful cases, cardiac catheterization may be of help In the presence of aortic insufficiency the pressure in the right ventricle and in the pulmonary artery will be normal or moderately elevated, secondarily to back pressure from the left side of the heart, whereas with 'primary pulmonary hypertension the pressure in the right ventricle and in the pulmonary artery will be markedly elevated

Aortic stenosis may be considered because of the attacks of syncope and precordial pain The two conditions are, however, readily differentiated, as aortic stenosis places a severe strain on the left ventricle, whereas in 'primary" pulmonary hypertension the strain falls on the right ventricle In the former the electrocardiogram shows evidence of extreme left ventricular hypertrophy, whereas in the latter there is extreme right ventricular hypertrophy

Polycythemia vera does not cause oxygen unsaturation of the arterial blood The electrocardiogram is usually normal, whereas electrocardiographic evidence of a right axis deviation and right ventricular hypertrophy is the rule in the malformation under discussion Furthermore, in polycythemia vera cardiac catheterization reveals a normal pressure in the pulmonary artery

✓*An Eisenmenger complex* differs from primary pulmonary hypertension in that a systolic murmur is more common and the second sound to the left of the sternum is not as forceful as in primary pulmonary hypertension The electrocardiogram shows greater evidence of left ventricular hypertrophy Fluoroscopy often reveals evidence of left auricular enlargement The circulation time is short There is oxygen unsaturation *of the arterial blood, which increases* with exercise Cardiac catheterization generally shows evidence of both a left to-right and a right to left shunt at the ventricular level

Ebstein's anomaly of the tricuspid valve can usually be differentiated from "primary" pulmonary hypertension in that the heart is larger and the heart

bed The increased resistance in the pulmonary vascular bed increases the work of the right side of the heart, there results right ventricular hypertrophy and dilatation of the main pulmonary artery and its branches

The course of the circulation is normal, except when the foramen ovale is held open by the high pressure in the right auricle and a right to-left shunt is thereby established

Dyspnea is the outstanding complaint

Engorgement of the liver and ascites develop as cardiac failure becomes severe

Syncope and precordial pain are late manifestations

The heart undergoes progressive enlargement, which is mainly right sided The pulmonic second sound is accentuated and there may be a long, loud, early diastolic murmur

Fluoroscopy reveals a fullness of the pulmonary conus, there may be a con spicuous hilar dance but the periphery of the lungs is clear

The electrocardiogram shows marked right axis deviation and evidence of right ventricular hypertrophy

The diagnosis is based upon the finding of a right sided cardiac enlargement and a markedly accentuated second sound over the pulmonary area The size of the heart, the age at which the patient develops symptoms, and the rapidity with which these advance to cardiac failure depend upon the etiology and the severity of the pulmonary vascular changes

The diagnosis is definitely established by cardiac catheterization and the find ing of a high pressure in the pulmonary artery, with a normal wedge pressure, and no evidence of intracardiac shunt

The malformation must be differentiated from other conditions which cause right sided cardiac strain with or without persistent cyanosis In early infancy the condition may be confused with a tetralogy of Fallot or a complete transposi tion of the great vessels As the heart enlarges, its contour becomes similar to that of a pure pulmonary stenosis In the absence of cyanosis, the condition is most commonly confused with idiopathic dilatation of the pulmonary artery, a corrected transposition of the great vessels, rheumatic or syphilitic heart disease with aortic insufficiency, and the Eisenmenger complex For a complete list see under Differential Diagnosis

Treatment is unsatisfactory but should be directed toward a reduction in the intrapulmonary pressure In the presence of cardiac failure digitalis is indicated

The prognosis is poor The pulmonary hypertension is usually progressive and eventually leads to cardiac failure and death

TREATMENT

At the present time treatment is only palliative Digitalis will help to strengthen the failing heart Any drugs[10] which offer the possibility of relaxation of the pulmonary vascular bed, such as priscoline or hexamethonium, may give some relief It is hoped that some drug may be discovered which will significantly reduce pulmonary pressure

The possibility of an allergic condition should be investigated As previously mentioned, the author has seen one patient who was greatly benefited by a summer in Maine, a part of the country which is known to be free from pollens and offers relief to many persons with hay fever Poppe, in discussion of a paper by Kaunitz and Andersen,[11] reported one patient in whom denervation of the lungs caused a long sustained reduction in the pulmonary pressure, but to the author s knowledge this is the only case in which denervation has been successful

There are probably many causes of "primary" pulmonary hypertension Nevertheless, until the etiology is known and a specific therapy is available, any measures which may reduce the pulmonary pressure should be given a trial

PROGNOSIS

The prognosis varies with the severity of the pulmonary hypertension It undoubtedly also varies with the etiology In very young patients in whom the pulmonary hypertension is more probably due to congenital abnormality of the pulmonary vascular bed, the slow expansion of the normal lung may compensate to a certain extent for the abnormality in the pulmonary vascular bed Consequently there may be considerable improvement during childhood A number of women have reached maturity and married and a few have been known to bear normal children Such, however, are the exception The condition generally progresses to an early fatal termination Death may result from cardiac failure or the circulation may become so sluggish that it is incompatible with life

SUMMARY

'Primary pulmonary hypertension means that there is increased resistance in the pulmonary vascular bed and that the heart itself is normally formed

The etiology is obscure and it is probable that there is more than a single etiological factor

"Primary" pulmonary hypertension may date from birth and there may be no evidence of infection demonstrated either during life or at autopsy, hence it is believed to be caused by a congenital abnormality in the pulmonary vascular

bed The increased resistance in the pulmonary vascular bed increases the work of the right side of the heart, there results right ventricular hypertrophy and dilatation of the main pulmonary artery and its branches

The course of the circulation is normal, except when the foramen ovale is held open by the high pressure in the right auricle and a right to-left shunt is thereby established

Dyspnea is the outstanding complaint

Engorgement of the liver and ascites develop as cardiac failure becomes severe

Syncope and precordial pain are late manifestations

The heart undergoes progressive enlargement, which is mainly right sided The pulmonic second sound is accentuated and there may be a long, loud, early diastolic murmur

Fluoroscopy reveals a fullness of the pulmonary conus, there may be a con spicuous hilar dance but the periphery of the lungs is clear

The electrocardiogram shows marked right axis deviation and evidence of right ventricular hypertrophy

The diagnosis is based upon the finding of a right sided cardiac enlargement and a markedly accentuated second sound over the pulmonary area The size of the heart, the age at which the patient develops symptoms, and the rapidity with which these advance to cardiac failure depend upon the etiology and the severity of the pulmonary vascular changes

The diagnosis is definitely established by cardiac catheterization and the find ing of a high pressure in the pulmonary artery, with a normal wedge pressure, and no evidence of intracardiac shunt.

The malformation must be differentiated from other conditions which cause right sided cardiac strain with or without persistent cyanosis In early infancy the condition may be confused with a tetralogy of Fallot or a complete transposi tion of the great vessels As the heart enlarges, its contour becomes similar to that of a pure pulmonary stenosis In the absence of cyanosis, the condition is most commonly confused with idiopathic dilatation of the pulmonary artery, a corrected transposition of the great vessels, rheumatic or syphilitic heart disease with aortic insufficiency, and the Eisenmenger complex For a complete list see under Differential Diagnosis

Treatment is unsatisfactory but should be directed toward a reduction in the intrapulmonary pressure In the presence of cardiac failure digitalis is indicated

The prognosis is poor The pulmonary hypertension is usually progressive and eventually leads to cardiac failure and death

TREATMENT

At the present time treatment is only palliative Digitalis will help to strengthen the failing heart Any drugs[10] which offer the possibility of relaxation of the pulmonary vascular bed, such as priscoline or hexamethonium, may give some relief It is hoped that some drug may be discovered which will significantly reduce pulmonary pressure

The possibility of an allergic condition should be investigated As previously mentioned, the author has seen one patient who was greatly benefited by a summer in Maine, a part of the country which is known to be free from pollens and offers relief to many persons with hay fever Poppe, in discussion of a paper by Kaunitz and Andersen,[11] reported one patient in whom denervation of the lungs caused a long sustained reduction in the pulmonary pressure, but to the author s knowledge this is the only case in which denervation has been successful

There are probably many causes of 'primary" pulmonary hypertension Nevertheless, until the etiology is known and a specific therapy is available, any measures which may reduce the pulmonary pressure should be given a trial

PROGNOSIS

The prognosis varies with the severity of the pulmonary hypertension It undoubtedly also varies with the etiology In very young patients in whom the pulmonary hypertension is more probably due to congenital abnormality of the pulmonary vascular bed, the slow expansion of the normal lung may compensate to a certain extent for the abnormality in the pulmonary vascular bed Consequently there may be considerable improvement during childhood A number of women have reached maturity and married and a few have been known to bear normal children Such, however, are the exception The condition generally progresses to an early fatal termination Death may result from cardiac failure or the circulation may become so sluggish that it is incompatible with life

SUMMARY

Primary" pulmonary hypertension means that there is increased resistance in the pulmonary vascular bed and that the heart itself is normally formed

The etiology is obscure and it is probable that there is more than a single etiological factor

'Primary' pulmonary hypertension may date from birth and there may be no evidence of infection demonstrated either during life or at autopsy, hence it is believed to be caused by a congenital abnormality in the pulmonary vascular

When the high pressure in the lesser circulation is due to changes in the pulmonary vascular bed and the heart itself is normally formed, the course of the circulation is as follows

The blood from the right auricle flows into the right ventricle and is pumped out into the pulmonary artery in the normal manner If the pressure in the pulmonary artery exceeds that in the descending aorta, a considerable volume of blood in the pulmonary artery will pass through the ductus arteriosus to the descending aorta The blood in the pulmonary artery which flows to the lungs is oxygenated in the lungs and returned in the normal manner to the left auricle, thence it flows into the left ventricle and is pumped out through the aorta to the systemic circulation Consequently the head and the upper extremities receive fully oxygenated blood from the left ventricle, whereas the trunk and lower extremities receive a mixture of fully oxygenated blood from the left ventricle and of venous blood which flows through the ductus arteriosus to the descending aorta It follows that the blood directed to the lower extremities is not fully oxygenated The blood from the upper extremities is returned by the superior vena cava to the right auricle and that from the lower extremities is returned by the inferior vena cava to the right auricle There the cycle starts again (see Diagram XVIII-3)

When the pulmonary hypertension is due to a severe malformation on the left side of the heart, the course of the circulation is altered only by the fact that a considerable part of the oxygenated blood returned to the left auricle will be shunted through the auricular defect into the right auricle, and consequently a mixture of oxygenated blood from the left auricle and venous blood from the right auricle will flow into the right ventricle and be pumped out through the pulmonary artery to the lungs and through the ductus arteriosus to the descending aorta Theoretically this means that the oxygen saturation of the blood in the descending aorta is not as low as when there is no shunt at the auricular level Nevertheless, inasmuch as the volume of the shunt varies with the severity of the pulmonary hypertension and the size of the ductus arteriosus, the oxygen unsaturation of the arterial blood in the descending aorta is subject to considerable variation Regardless of the presence or absence of an auricular defect and a malformation in the left side of the heart, the head and the upper extremities receive fully oxygenated blood which has been pumped from the left ventricle into the aorta and in both instances the trunk and lower extremities receive some venous blood (see Diagram XVIII-4)

B *Pulmonary Hypertension with Persistent Patency of the Ductus Arteriosus*

Pulmonary hypertension combined with persistent patency of the ductus arteriosus produces a distinctive clinical syndrome when the pressure in the pulmonary artery is greater than the systemic pressure

NATURE OF THE MALFORMATION

The essential feature of this malformation is the combination of severe pulmonary hypertension and persistent patency of the ductus arteriosus Consequently any condition which causes the pulmonary pressure to be higher than the systemic pressure, when it occurs in combination with persistent patency of the ductus arteriosus, will produce this clinical syndrome [1-13] The pulmonary hypertension may be primary, that is, due to increased resistance in the pulmonary vascular bed, or it may be secondary to a severe malformation on the left side of the heart combined with a gross defect in the auricular septum

The malformation may be that of severe mitral stenosis or there may be defective development of the left ventricle and marked hypoplasia of the ascending aorta In either instance, if there is also a gross defect in the auricular septum, as soon as the pressure in the left auricle exceeds that in the right auricle, a considerable volume of the blood returned to the left auricle will be shunted into the right auricle and thence to the right ventricle The mixture of oxygenated blood from the left auricle and of venous blood from the right auricle, which flows into the right ventricle, will be pumped out through the pulmonary artery to the lungs and through the ductus arteriosus to the descending aorta

It is conceivable, although still unproven, that an excessively large ductus arteriosus in which the pulmonary hypertension is initially compensatory may ultimately lead to irreversible changes in the lungs which will progress to such a point that the pressure in the pulmonary artery will exceed systemic pressure Consequently the flow of blood through the ductus arteriosus will again be reversed The author believes that this *may* occur when a child with an enormous ductus arteriosus regains compensation and is extremely active It is, however, the exception, not the rule

COURSE OF THE CIRCULATION

The course of the circulation varies with the nature of the pulmonary hypertension

DIAGRAM XVIII–3

Primary pulmonary hypertension with patency of the ductus arteriosus

In this malformation there is increased resistance in the pulmonary vascular bed and the ductus arteriosus is patent.

The blood from the right auricle flows into the right ventricle and is pumped into the pulmonary artery Part of the blood in the pulmonary artery flows to the lungs and, because of the high pressure in the pulmonary artery part flows through the ductus arteriosus into the descending aorta The blood which flows to the lungs is oxygenated and is returned in the normal fashion to the left auricle Thence it flows to the left ventricle and is pumped out through the aorta to the systemic circulation The blood which flows to the head and the upper extremities is returned in the normal manner by the superior vena cava to the right auricle The blood which flows through the arch of the aorta to the descending aorta meets the venous blood which flows through the ductus arteriosus to the descending aorta This mixture of arterial and venous blood flows to the trunk and the lower extremities and is returned by the inferior vena cava to the right auricle. There the cycle starts again

Clinical diagnosis is based upon the fact that the blood which flows to the lower extremities is not fully saturated hence the feet are cyanotic and the head and the upper extremities of normal color The heart is normal in size The pulmonic second sound is accentuated and there may be marked pulmonary insufficiency The electrocardiogram shows a right axis deviation and evidence of right ventricular hypertrophy

Diagram XVIII–3

Head and upper extremities

Ductus Arteriosus

R Lung

P.A.

S.V.C.

Aorta

P.A.

L Lung

PV

PV

Pulmonary Artery

LA

R.A.

LV

R.V.

Descending Aorta

I.V.C.

Trunk and lower extremities



Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

DIAGRAM XVIII-4

Primary pulmonary hypertension combined with a severe left sided heart lesion and persistent patency of the ductus arteriosus

In this malformation there is a gross abnormality on the left side of the heart either mitral stenosis or underdevelopment of the left ventricle and hypoplasia of the ascending aorta, or some other severe left sided cardiac lesion combined with a gross defect in the auricular septum

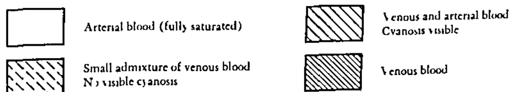
Under such circumstances the blood from the right auricle flows into the right ventricle and is pumped out through the pulmonary artery to the lungs and also through the ductus arteriosus to the descending aorta The blood which flows to the lungs is fully oxygenated and is returned in the normal manner to the left auricle. Owing to the malformation on the left side of the heart, it is difficult for the blood in the left auricle to flow into the left ventricle and to be pumped out through the aorta to the systemic circulation. Nevertheless the blood which does flow to the left ventricle and thence to the systemic circulation is fully oxygenated The high pressure in the left auricle combined with the defect in the auricular septum, means that a considerable volume of oxygenated blood is shunted from the left auricle to the right auricle, where it mixes with the venous blood returned by the superior vena cava and the inferior vena cava to that chamber This mixture of oxygenated and venous blood flows into the right ventricle So the cycle continues

In this combination of anomalies two factors increase the pressure in the pulmonary artery The right ventricle pumps blood through the ductus arteriosus to the descending aorta hence the pressure in the pulmonary artery is the same as that in the descending aorta In addition the high pressure in the left auricle elevates the pressure in the pulmonary capillary bed Consequently there is severe pulmonary hypertension If the pressure in the descending aorta is significantly lower than that in the pulmonary artery as so frequently occurs with a severe left sided lesion some venous blood in the descending aorta may be forced back into the left subclavian artery, causing cyanosis of the left hand as well as of the feet.

Clinical diagnosis The difference in cyanosis between the two hands or between the hands and the feet, permits the diagnosis to be made at sight Although special studies may be necessary to determine the existence of a malformation on the left side of the heart, such a lesion should be suspected if the left hand is more cyanotic than the right hand

Diagram XVIII–4

Head and upper extremities
Ductus Arteriosus
R Lung
R.A.
S.V.C.
Aorta
Pul. A
L Lung
PV
Pulmonary Artery
Pul.V
L.A
Descending
R.A
Mitral stenosis
L V
R.V
I.V.C.
Aorta
Trunk and lower extremities



artery may be so great that there is clubbing of the fingers of the left hand but not of the right Under such circumstances the superficial appearance is that of unilateral cyanosis namely, the left side of the body appears to be cyanotic and the right side of normal color Usually careful examination of the back will show that the head and neck and the tops of the shoulders are of normal color and the remainder of the thorax is cyanotic The line of demarcation of the cyanosis lies at a slightly lower level on the right shoulder than on the left, as the upper part of the right shoulder receives its blood supply from the right scapular artery, which carries fully oxygenated blood

The difference in cyanosis between the upper and lower extremities is usually sufficiently marked so that the diagnosis can be made at sight

Polycythemia develops slowly Initially the patient is not polycythemic and may even be anemic In the presence of anemia the volume of reduced hemoglobin circulating through the lower extremities may be too slight to produce 'visible' cyanosis As the polycythemia increases, the patient has the high color and the plethoric appearance so commonly seen in polycythemia vera

Difficulty in feeding and *failure to gain* are common complaints in early infancy

Repeated pulmonary infections are of such frequent occurrence that it sometimes seems as if the infant has hardly recovered from one infection before he develops another

Dyspnea on exertion is also a common complaint It varies with the severity of the pulmonary hypertension It is usually the development of exertional dyspnea which brings the patient to the doctor Thereafter dyspnea persists as the outstanding difficulty and becomes increasingly severe throughout the remainder of the patient's life

CARDIAC FINDINGS

The size of the heart varies with the underlying pathology If the condition is one of 'primary' pulmonary hypertension combined with patency of the ductus arteriosus the heart may be remarkably small, because the ductus arteriosus acts as an escape valve for the high pressure in the pulmonary artery (see Figure XVIII-4) Indeed the patency of the ductus arteriosus means that the pressure in the pulmonary artery cannot greatly exceed that in the systemic circulation and thus in turn limits the work required of the heart Nevertheless, the right ventricle becomes hypertrophied and the pulmonary artery and its main branches are dilated consequently the pulmonary conus is prominent *The pul*

PHYSIOLOGY OF THE MALFORMATION

When the ductus arteriosus remains patent, as soon as the pressure in the pulmonary artery exceeds systemic pressure, the direction of the flow of blood through the ductus will be reversed, that is, blood will flow from the pulmonary artery into the descending aorta For this reason the condition is frequently called a 'reversed ductus" When this occurs, the ductus arteriosus acts as an escape valve, it prevents the pressure in the pulmonary artery from ever greatly exceeding systemic pressure and thereby lessens the load placed upon the heart It does not, however, limit the extent of injury to the lungs There is progressive narrowing of the pulmonary vascular bed which causes an ever increasing volume of blood to be shunted through the ductus arteriosus into the descending aorta

If there is a severe lesion on the left side of the heart, the pressure in the systemic circulation is frequently abnormally low Hence there is a greater tendency for the pressure in the pulmonary artery to be significantly higher than that in the descending aorta There is a correspondingly greater likelihood for some of the blood which is shunted from the pulmonary artery through the ductus arteriosus to the descending aorta to be directed back into the left subclavian artery Under such circumstances the left hand may appear slightly cyanotic It follows that, when the left hand is more cyanotic than the right, the chances are in favor of the existence of a malformation on the left side of the heart

CLINICAL FINDINGS

The distribution of the cyanosis gives the clue to the diagnosis The head and the upper extremities are of normal color, whereas the lower extremities, which receive the mixture of oxygenated blood from the ascending aorta and of venous blood from the pulmonary artery, are cyanotic When the pressure in the aorta is lower than that in the pulmonary artery, owing to the close proximity of the left subclavian artery to the point of entrance of the ductus arteriosus, some venous blood is forced back up the aorta and not infrequently the left subclavian artery receives some venous blood, together with the fully oxygenated blood from the ascending aorta Consequently the left hand may be more cyanotic than the right Often the difference in cyanosis between the two hands is not great, even the difference in cyanosis between the left hand and the feet may not be convincing but, upon comparison of the right hand with either foot, the difference in the color is readily apparent

At times the oxygen unsaturation of the arterial blood in the left subclavian

X RAY AND FLUOROSCOPIC FINDINGS

When primary pulmonary hypertension is associated with persistent patency of the ductus arteriosus, the heart is usually normal in size but, when the condition is associated with a malformation on the left side of the heart, there may be considerable cardiac enlargement In the anterior posterior position the pulmonary conus is prominent and the main branches of the pulmonary artery are slightly dilated (see Figure xviii-4) Sometimes there is enormous dilatation of the pulmonary artery and its main branches, as shown in Figure xviii-5, in this instance the condition had progressed so far that there was calcification in the main branches of the pulmonary artery In the left anterior-oblique position the right ventricle usually appears to be slightly to moderately enlarged The right anterior-oblique position is usually best for the determination of left auricular enlargement If the left auricle is enlarged, it is strong evidence that the pulmonary hypertension is secondary to a serious abnormality of the left auricle or the left ventricle and that the defect in the auricular septum is small

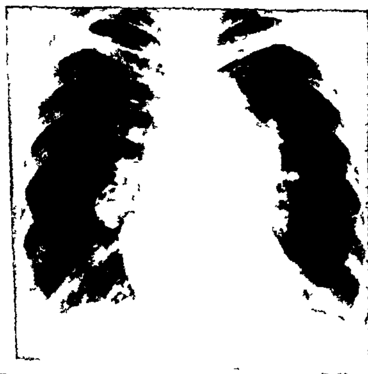


Figure xviii-5 "Primary pulmonary hypertension and patency of the ductus arteriosus Adult

Note calcification in the aorta and the main branches of the pulmonary artery



FIGURE XVIII-4 Primary' pulmonary hypertension and patency of the ductus arteriosus Adult

monic second sound is accentuated and usually reduplicated There may be a *long, loud early diastolic murmur* along the left sternal border due to pulmonary insufficiency

When the pulmonary hypertension is secondary to a malformation of the left auricle or the left ventricle, the heart may or may not be enlarged Furthermore, there may or may not be clinical evidence of mitral stenosis In some in stances a presystolic murmur may be audible at the apex In other instances a severe malformation of the left side of the heart may produce no abnormal mur mur or thrill A severe malformation on the left side of the heart frequently causes the pressure in the systemic circulation to be abnormally low

X RAY AND FLUOROSCOPIC FINDINGS

When primary pulmonary hypertension is associated with persistent patency of the ductus arteriosus, the heart is usually normal in size but, when the condition is associated with a malformation on the left side of the heart, there may be considerable cardiac enlargement In the anterior posterior position the pulmonary conus is prominent and the main branches of the pulmonary artery are slightly dilated (see Figure xviii–4) Sometimes there is enormous dilatation of the pulmonary artery and its main branches, as shown in Figure xviii–5, in this instance the condition had progressed so far that there was calcification in the main branches of the pulmonary artery In the left anterior-oblique position the right ventricle usually appears to be slightly to moderately enlarged The right anterior-oblique position is usually best for the determination of left auricular enlargement If the left auricle is enlarged, it is strong evidence that the pulmonary hypertension is secondary to a serious abnormality of the left auricle or the left ventricle and that the defect in the auricular septum is small

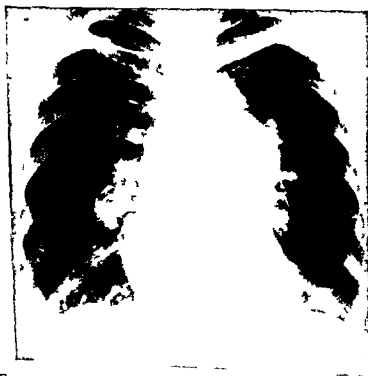


Figure xviii–5 Primary pulmonary hypertension and patency of the ductus arteriosus Adult

Note calcification in the aorta and the main branches of the pulmonary artery



FIGURE XVIII–4 Primary ' pulmonary hypertension and patency of the ductus arteriosus Adult

monic second sound is accentuated and usually reduplicated There may be a *long, loud early diastolic murmur* along the left sternal border due to pulmonary insufficiency

When the pulmonary hypertension is secondary to a malformation of the left auricle or the left ventricle, the heart may or may not be enlarged Further more, there may or may not be clinical evidence of mitral stenosis In some in stances a presystolic murmur may be audible at the apex In other instances a severe malformation of the left side of the heart may produce no abnormal mur mur or thrill A severe malformation on the left side of the heart frequently causes the pressure in the systemic circulation to be abnormally low

Cardiac catheterization will reveal high pressure in the right ventricle and in the pulmonary artery Since the direction of the flow of blood is through the ductus arteriosus to the descending aorta, it is frequently possible to pass the catheter into the descending aorta Cardiac catheterization, however, gives little or no information concerning the presence or absence of a malformation on the left side of the heart

Angiocardiography is less dangerous in this combination of anomalies than when the ductus arteriosus is closed, as the ductus arteriosus acts as an escape valve and thus prevents the accumulation of dye in the lungs, furthermore, the brain is supplied with fully oxygenated blood

Angiocardiography will reveal that the descending aorta fills simultaneously with the pulmonary artery In patients with mitral stenosis or marked hypoplasia of the mitral valve, the dye will linger for a long time in the left auricle, the left ventricle and ascending aorta may never be visualized

Aortography may be helpful in the determination of the size of the ascending aorta and the exclusion of a coarctation of the aorta In order to obtain a satisfactory aortogram, the dye must be injected into an artery on the same side of the body as that toward which the aorta arches that is, with a normal left aortic arch the test should be performed through the left brachial artery If the dye is injected on the opposite side to that of the descending aorta, the increased pressure transmitted from the pulmonary artery to the descending aorta will lessen the flow of blood from the ascending aorta to the descending aorta, consequently the dye may pass only up the carotid arteries to the head This is especially prone to occur if the left hand is cyanotic Under such circumstances the aortogram may simulate that of a complete interruption of the aortic arch even though in reality the arch is intact

When aortography is properly performed, it is of great diagnostic aid The arch of the aorta and the ductus arteriosus may both be visualized The contrast media enters the ductus and delineates it but, owing to the high pressure in the pulmonary artery, no dye enters the lungs This finding is in striking contrast to the simultaneous visualization of the aorta and the pulmonary arteries in an infant with a large ductus arteriosus and pulmonary hypertension when the predominant shunt is from the aorta to the lungs (see Chapter xx)

DIAGNOSIS

The diagnosis may be made at sight when the left hand is cyanotic and the right hand is of normal color The diagnosis can also be made at sight when the face and the upper extremities are of normal color and the feet are cyanotic

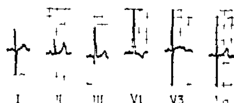


Figure XVIII–6 Primary pulmonary hypertension and patency of the ductus arteriosus

Fluoroscopy frequently reveals pulsations in the main pulmonary artery but, owing to the fact that the ductus arteriosus acts as an escape valve, these pulsations are generally minimal and do not extend into the small branches of the pulmonary artery The periphery of the lungs is usually abnormally clear

ELECTROCARDIOGRAPHIC FINDINGS

The standard leads show a right axis deviation and the unipolar precordial leads show evidence of marked right ventricular hypertrophy (see Figure XVIII–6) Inasmuch as the patency of the ductus arteriosus relieves the strain on the right ventricle, the evidence of right ventricular hypertrophy is frequently not as marked as when primary pulmonary hypertension occurs as an isolated malformation Indeed, the author has seen one patient in whom the lesion on the left side of the heart was such as to give a left axis deviation, but even in that instance the precordial leads showed evidence of right ventricular hypertrophy

SPECIAL TESTS

A difference in the oxygen saturation of arterial blood between the right brachial artery and the femoral artery is characteristic of this malformation, because the head and the upper extremities receive fully oxygenated blood from the ascending aorta and the femoral artery receives some venous blood which is directed through the ductus arteriosus to the descending aorta In order to be certain that the difference between the right brachial artery and the femoral artery is significant, the two blood samples must be drawn simultaneously It is, of course, unfortunate but inevitable that the sample of blood from the femoral artery will approximate that of normal venous blood If, however, a sample of venous blood is drawn, the oxygen content of the venous blood will be even lower than that of the femoral arterial blood Furthermore, the pressure in the femoral artery is always higher than that in the femoral vein

Additional studies are usually necessary to determine whether the pulmonary hypertension is primary or secondary, that is, whether the principal difficulty occurs in the lungs or in the heart Such knowledge is obviously essential as a guide to therapy

ondary to the large ductus arteriosus or whether the patency of the ductus arteriosus is secondary to the pulmonary hypertension

Pulmonary hypertension secondary to the increased circulation to the lungs is the rule in infants with a large ductus arteriosus Such *infants* show great cardiac enlargement, a systolic and a mid-diastolic murmur over the precordium, enlargement of the left auricle, increased vascular shadows, and electrocardiographic evidence of 'combined hypertrophy

In contrast to these findings, when the pulmonary hypertension is primary and the ductus arteriosus has remained patent, the patient shows cyanosis of the *lower extremities*, the heart is usually of normal size, there may or may not be a systolic murmur, the pulmonary vascularity is reduced, and the electrocardiogram shows evidence of right ventricular hypertrophy

An aortogram sharply differentiates the two conditions When the pulmonary hypertension is secondary to the flow of blood through the ductus arteriosus, dye is seen in the lungs as the aorta is visualized, whereas when the pulmonary hypertension is primary, although the ductus arteriosus may be opacified, no dye enters the lungs

Cardiac catheterization will aid in the differentiation of the two conditions When the pulmonary hypertension is secondary to a large ductus, it is the large left to-right shunt which causes the hypertension, under such circumstances there is a marked increase in the oxygen content of the blood in the pulmonary artery On the other hand, when the pulmonary hypertension is primary, the shunt is from right to-left, there is little or no evidence of a left to-right shunt, and no increase in the oxygen content of the blood in the pulmonary artery

Thus the extreme of the one is readily differentiated from the extreme of the other In early infancy the two conditions are usually readily differentiated In older patients there may be real difficulty in the differentiation of pulmonary hypertension combined with persistent patency of the ductus arteriosus from a patent ductus arteriosus which causes pulmonary hypertension (see Chapter XX)

If the flow of blood from the pulmonary artery to the aorta is sufficient to cause cyanosis of the lower extremities, or even if there is a clearly detectable difference in the oxygen saturation of the arterial blood in the brachial and femoral arteries the pulmonary hypertension has *progressed* to the point where it, not the ductus arteriosus, is the primary difficulty Under such circumstances, unless the pressure in the pulmonary artery can be relieved, ligation of the ductus arteriosus is usually contraindicated

Complete transposition of the great vessels combined with a patency of the

Sometimes the history of a slight cardiac disability throughout life, combined with the finding of a loud diastolic murmur, will direct attention to the possibility of pulmonary hypertension and will lead to careful examination of the color of the right hand and of the feet

The determination of the oxygen saturation of the arterial blood samples drawn simultaneously from the right arm and from either of the femoral arteries clinches the diagnosis

Unless there is frank evidence of mitral stenosis or evidence of left auricular enlargement, further studies are usually necessary to determine whether the pulmonary hypertension is primary or secondary to a severe malformation on the left side of the heart

DIFFERENTIAL DIAGNOSIS

This condition requires differentiation from 'primary" pulmonary hypertension (see Section A) and also from other conditions which cause a difference in cyanosis between the upper and the lower extremities, such as coarctation of the aorta with the ductus arteriosus opening below the constriction, patency of the ductus arteriosus associated with pulmonary hypertension, complete transposition of the great vessels, and complete interruption of the isthmus of the aorta when the descending aorta is continuous with the pulmonary artery through the ductus arteriosus

Coarctation of the aorta with the ductus arteriosus opening below the constriction theoretically would give the same clinical picture as a pulmonary hypertension with a persistent patency of the ductus arteriosus An infant with a coarctation of the aorta frequently develops severe right sided cardiac failure, furthermore, as the ductus periodically clamps down, no pulse is palpable in the femoral artery Aortography performed through the left subclavian artery will readily show whether or not there is a coarctation of the aorta

If there is cyanosis of the left hand, it is clear evidence that there is reflux of blood from the pulmonary artery into the left subclavian artery, hence there cannot be a constriction of the aorta between the left subclavian artery and the point of entrance of the ductus arteriosus

Occasionally a large patent ductus arteriosus presses upon and constricts the aorta and thus causes a functional coarctation of the aorta An aortogram usually reveals the true nature of the difficulty

A large patent ductus arteriosus associated with pulmonary hypertension presents a diagnostic problem which is both extremely difficult and important Almost invariably the question arises whether the pulmonary hypertension is sec

Coarctation of the aorta can be corrected by surgery In young infants who suffer from severe cardiac failure, it is, however, usually preferable to close the ductus and at a later date to correct the coarctation (see Chapter XXVII)

If the pulmonary hypertension is secondary to severe mitral stenosis, early relief of the mitral stenosis may be of benefit When the mitral stenosis is due to rheumatic fever, operation is relatively simple, but when the mitral stenosis is *congenital*, the problem is much more difficult (see Chapter XXX) All too frequently the mitral valve and the left auricle may be so grossly abnormal that it is impossible to improve the circulation Dr Clifford Parsons[14] has reported one successful operation on a congenital mitral valve, but in this case the operation did not alter the pulmonary pressure

Complete interruption of the isthmus of the aorta offers a challenge to the surgeon to reunite the proximal and distal ends of the aorta, either directly or by graft, and then to divide the ductus arteriosus Unfortunately, however, the condition is usually associated with additional cardiac abnormalities which must also be corrected in order to help the infant

PROGNOSIS

The prognosis varies with the etiology and the severity of the pulmonary hypertension By and large the prognosis is better when 'primary" pulmonary hypertension is combined with patency of the ductus arteriosus than when the ductus undergoes normal obliteration This is an additional reason for the author s great hesitancy in recommending surgical closure of the ductus unless the ductus arteriosus is clearly the cause of the hypertension The author has known several women in their early twenties who were only slightly incapacitated by this condition, one woman has married and has had one normal pregnancy without great cardiac difficulty Thus, although the condition is extremely serious, it may be compatible with life for a number of years

SUMMARY

Pulmonary hypertension combined with persistent patency of the ductus arteriosus produces a distinctive clinical syndrome

The pulmonary hypertension may be primary or may be secondary to a severe malformation on the left side of the heart In either case, owing to the high pressure in the lungs, blood flows from the pulmonary artery through the ductus arteriosus to the descending aorta Consequently the head and the upper extremities receive fully oxygenated blood and the trunk and the lower extremities receive a mixture of venous and arterial blood Therefore the head and

ductus arteriosus may be confused with the malformation under discussion be cause of the difference in cyanosis between the upper and the lower extremities The difference in cyanosis in a complete transposition of the great vessels is the reverse of that which occurs in primary pulmonary hypertension and persistent patency of the ductus arteriosus that is, the head and the upper extremities are more cyanotic than are the trunk and the lower extremities Moreover, when there is a complete transposition of the great vessels, inasmuch as the pressure in the aorta is normal and that in the pulmonary artery is so high that blood flows from the pulmonary artery into the descending aorta, the line of demarca tion of the cyanosis lies at the brim of the pelvis In contrast to this, when the pressure in the systemic circulation is so low that the systemic pressure is main tained by the flow of blood from the pulmonary artery through the ductus arteriosus to the aorta, the line of demarcation of the cyanosis lies high up on the shoulder girdle

Complete interruption of the isthmus of the aorta may also be confused with this malformation An aortogram, properly taken, should enable the physi cian to make the correct diagnosis

TREATMENT

The treatment obviously depends upon the cause of the pulmonary hyperten sion

If the hypertension is secondary to a left sided cardiac lesion, attention should be directed to the correction of the cardiac anomaly

The crucial question is whether the pulmonary hypertension is secondary to the patency of the ductus arteriosus or the pulmonary hypertension is primary and the ductus arteriosus acts as an escape valve to relieve the high pressure in the lungs

The differentiation of these two conditions is extremely important because, if the ductus arteriosus is the cause of the pulmonary hypertension, ligation of the ductus restores the heart and circulation to normal (see Chapter xx), whereas, if the pulmonary hypertension is primary, ligation of the ductus ar teriosus usually causes great dilatation of the right side of the heart and may lead to cessation of heart action Even in instances in which the patient survives the closure of the ductus, the benefit derived therefrom is doubtful Indeed, closure of the ductus arteriosus may ultimately do harm because it closes the escape valve It is for these reasons that the author is strongly opposed to closure of the ductus arteriosus in a patient with severe pulmonary hypertension unless the pulmonary pressure can be reduced

the ostium infundibuli associated with biventricular origin of the aorta Circulation 2 545-552, 1950
4 Edwards J E, J M Douglas, H B Burchell, and N A. Christensen Pathology of the intrapulmonary arteries and arterioles in coarctation of the aorta associated with patent ductus arteriosus Am Heart J 38 205-233, 1949
5 Dresdale, D T., M Schultz, and R. J Michtom Primary pulmonary hypertension clinical and hemodynamic study Am J Med 11 686-705 1951
6 Coleman P N., A W B Edmunds, and J Tregillus Primary pulmonary hypertension in 3 sibs Brit. Heart J 21 81-88, 1959
7 Rich A R. A hitherto unrecognized tendency to the development of widespread pulmonary vascular obstruction in patients with congenital pulmonary stenosis (tetralogy of Fallot) Bull Johns Hopkins Hosp 82 389-401 1948
8 Lilienthal J L., Jr and R. L. Riley Diseases of the respiratory system, circulation through the lung and diffusion of gases. Ann. Rev Med. 5 237-284, 1954
9 Harvey J C Myxoma of the left auricle. Ann. Int Med 47 1067-1081, 1957
10 Dresdale, D T, R J Michtom, and M Schultz. Recent studies in primary pulmonary hypertension including pharmacodynamic observations on pulmonary vascular resistance. Bull. New York Acad Med 30 195-207, 1954
11 Kaunitz, V H and M. N Andersen An experimental study of the effect of parasympathetic denervation of the lung on pulmonary artery pressure (see discussion by J K. Poppe) J Thoracic Surg 27 55-63 1954
12 Hultgren H A Selzer A. Purdy E Holman and F Gerbode. The syndrome of patent ductus arteriosus with pulmonary hypertension. Circulation 8 15-35, 1953
13 Taussig H B R S Bauersfeld and A J MacDonald Pulmonary hypertension with persistent patency of the ductus arteriosus Presented at American Pediatric Society Meeting Old Point Comfort, Virginia May 1952
14 Parsons C. Personal communication

the upper extremities are of normal color and the trunk and the lower extremities are cyanotic The distribution of the cyanosis permits diagnosis at sight

The cardiac findings vary with the etiology of the hypertension If the pulmonary hypertension is primary, the heart is usually relatively small The second sound over the pulmonary area is accentuated and frequently there is a loud diastolic murmur audible along the left sternal border

X ray and fluoroscopy show that the heart is not greatly enlarged, there may be a minimal hilar dance The periphery of the lungs is abnormally clear If the pulmonary hypertension is secondary to a malformation of the left auricle or the left ventricle, x ray and fluoroscopy may reveal evidence of left auricular enlargement

The electrocardiogram usually shows right axis deviation and evidence of right ventricular hypertrophy

The determination of the oxygen saturation of samples of arterial blood taken simultaneously from the right brachial artery and either femoral artery will clinch the diagnosis

Although diagnosis may be made at sight, special studies are usually necessary to determine the nature of the pulmonary hypertension

Angiocardiography is of greater aid than cardiac catheterization It demonstrates the flow of blood from the pulmonary artery to the descending aorta and it also gives some information concerning the left side of the heart

The condition requires differentiation from coarctation of the aorta combined with patency of the ductus arteriosus, from simple patency of a large ductus arteriosus in which there is pulmonary hypertension, and also from complete interruption of the isthmus of the aorta in which the descending aorta is continuous with the pulmonary artery through the ductus arteriosus

The treatment varies with the etiology

Prognosis is guarded but the condition may be compatible with life for a number of years

References

1 Edwards, J E Structural changes of the pulmonary vascular bed and their functional significance in congenital cardiac disease, 26th Ludvig Hektoen Lecture Proc Inst Med Chicago 18 134–146, 1950

2 Civin, H W and J E Edwards The postnatal structural changes in the intrapulmonary arteries and arterioles A M A Arch Path 51 192–200, 1951

3 Civin, H W and J E Edwards Pathology of the pulmonary vascular tree, a comparison of the intrapulmonary arteries in the Eisenmenger complex and in stenosis of

NATURE OF THE MALFORMATION

The essential feature of the malformation concerns the anomaly of the tri cuspid valve and of the upper portion of the wall of the right ventricle One or more leaflets of the tricuspid valve are fused with the ventricular endocardium over large areas Consequently it is often difficult to identify the individual leaf lets with certainty The anterior leaflet is the least involved, indeed, it is often normal The medial or septal leaflet may or may not be normal Frequently it is sealed to the septal wall and in some instances it is entirely absent The posterior leaflet is usually the most malformed and is displaced down toward the apex of the right ventricle In most instances the upper portions of the leaflet are blended with the endocardium of the right ventricle and the distal margins of the valve which are free become fused together to form a basket network deep within the right ventricle Consequently there is no valve at the normal tricuspid orifice but a new valve is formed by the basket network Sometimes this new valve has a normal orifice which opens into the lower portion of the right ventricle In other instances there is no true orifice and the blood passes through the interstices of the basket network into the outflow tract of the right ventricle In either event the new valve which is formed deep within the right ventricle divides this cham ber into two parts That portion of the right ventricle which lies above the valve becomes incorporated into the right auricle and only that portion of the right ventricle which lies below the valve is of functional importance in the expulsion of blood to the lungs

The extent to which the tricuspid valve is plastered against the right ventricle and the position of the new valve vary from case to case In some instances the abnormality is relatively slight and the position of the new valve is such that the lower chamber is the larger of the two cavities Such is the anomaly shown in Figure XIX-1 In other instances virtually the entire tricuspid valve is plastered against the endocardium of the right ventricle and the new valve extends nearly to the apex of the right ventricle Under such circumstances the lower chamber, which is the outflow tract of the right ventricle, is abnormally small, as illustrated in Figures XIX-2 and 3 Nevertheless, the tricuspid valve may be abnormally large and during ventricular systole may balloon upward into the right auricle, thus increasing the size of the lower chamber This chamber is, however, usually too small to receive all the blood which has been returned to the right auricle and to pump the blood through the pulmonary artery to the lungs

Engle and her a sociates[3] have pointed out that the extreme thinness of the

CHAPTER XIX

EBSTEIN'S ANOMALY

CONGENITAL DOWNWARD DISPLACEMENT OF THE TRICUSPID VALVE

IN 1866 Ebstein[1] first reported the malformation which now bears his name The anomaly consists of a congenital downward displacement of the tricuspid valve into the right ventricle The tricuspid valve is distorted in such a manner that one or more of its leaflets become plastered against the wall of the right ventricle, consequently there is no valve at the annulus fibrosus which separates the right auricle from the right ventricle For this reason Ebstein described the abnormality as a severe degree of congenital insufficiency of the tricuspid valve Actually in most instances a new valve is formed deep within the right ventricle, usually this valve is functionally sufficient The right ventricular wall above the anomalous valve is extremely thin The anomaly has been reviewed by Yater and Shapiro[2] and the clinical syndrome has been clarified by Engle et al[3] and by Schiebler et al[4]

EMBRYOLOGY

A brief review of the early embryology of the musculature of the heart will aid in understanding the nature of the malformation Streeter[5] has shown that the myocardium arises from a specialized portion of the visceral celomic wall and is separated from the endocardium of the primitive heart by the myoendocardial space which is filled with a homogenous transparent material known as the cardiac jelly' As the myocardium develops, this jelly gradually disappears, except at strategic sites, where it persists to form the so-called endocardial cushions which are the precursors of the valves Thus, as Engle et al[3] have postulated, 'any defect in the visceral celomic wall in the region where the right ventricle is to develop could not only cause defective development of the right ventricular myocardium but also by distortion of the position of the persisting primitive myoendocardial space could cause a malformation of the tricuspid valve" Be that as it may, defective development of the wall of the right ventricle above the anomalous tricuspid valve appears to be an integral part of the malformation

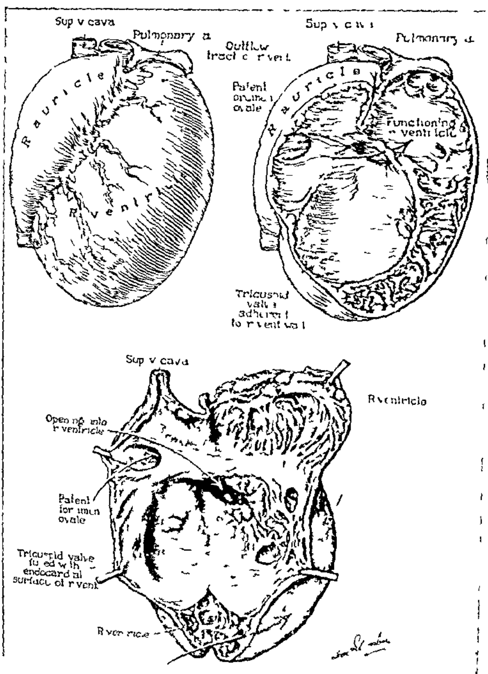


FIGURE XIX-2 Ebstein s anomaly with marked displacement of the tricuspid valve (same patient as in Figures XIX-5 7) Child

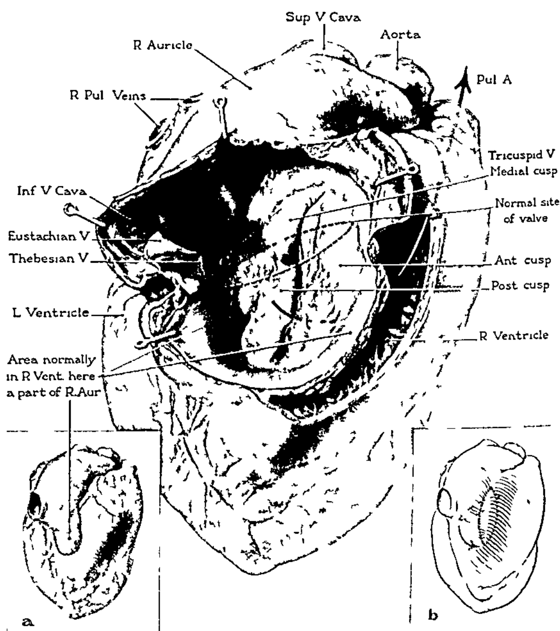


FIGURE XIX-1 Ebstein's anomaly with relatively slight displacement of the tricuspid valve (same patient as in Figure XIX-4) Young adult

myocardium in the upper portion of the right ventricular wall which lies above the new valve is an integral part of the malformation The myocardium in this region may be only 1 or 2 mm in thickness Localized aneurysmal dilatation of the right ventricular wall in this area has been reported by Blackhall Morison[1] and Brekke[7] In each instance the aneurysm was believed to be due to congenital weakness of the wall That portion of the wall of the right ventricle which lies below the valve is of normal thickness but the lower cavity is abnormally small, the ventricular septum is intact

In addition to the malformation of the tricuspid valve, there is frequently some abnormality either of the eustachian valves or of the thebesian valves Such anomalies are, however, only of academic interest

The foramen ovale may or may not be patent Although in some instances the foramen ovale closes in the normal manner and becomes completely sealed, it remains anatomically and functionally patent in two-thirds of the patients with this malformation If the valve covering the foramen ovale is not completely sealed, the high pressure in the right auricle will force the valve to open Indeed, the increased pressure in the right auricle caused by the misplaced tricuspid valve probably explains the high incidence of patency of the foramen ovale which occurs in association with this malformation[3] When the foramen ovale remains patent it acts as an escape valve for the high pressure in the right auricle Thereby it becomes of functional importance and a right to-left shunt is established

COURSE OF THE CIRCULATION

The course of the circulation depends upon whether or not the valve covering the foramen ovale becomes completely sealed So long as the foramen ovale is closed and in all instances in which the foramen ovale becomes completely sealed the circulation of the blood is normal Under such circumstances the blood from the right auricle flows into the right ventricle and is pumped out into the pulmonary artery The blood in the pulmonary artery is directed to the lungs where it is oxygenated, and is returned in the normal fashion by the pulmonary veins to the left auricle and thence to the left ventricle The blood in the left ventricle is pumped out by way of the aorta to the systemic circulation and is returned in the normal manner by the superior vena cava and the inferior vena cava to the right auricle (see Diagram XIX-1) Inasmuch as there is no communication between the two sides of the heart, there is no venous-arterial shunt Hence there is no cyanosis and no clubbing

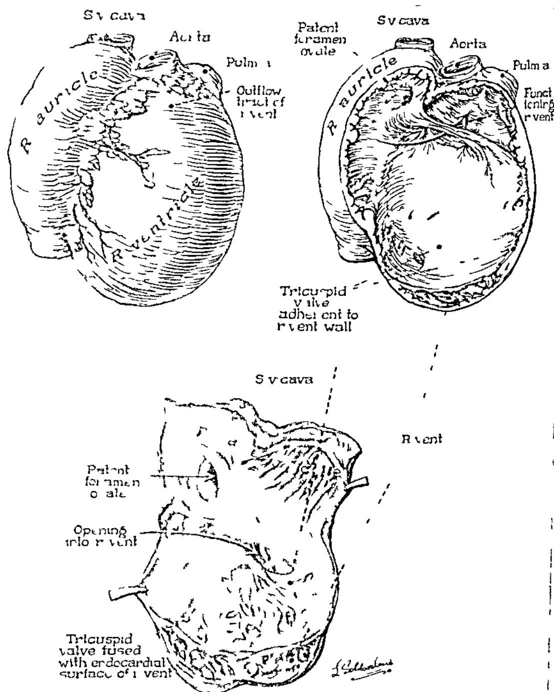


FIGURE XIX-5 Ebstein's anomaly with extreme displacement of the tricuspid valve (same patient as in Figure XIX-6) Young child

DIAGRAM XIX–1

Ebstein's anomaly of the tricuspid valve

The essential feature of this malformation is a congenital downward displacement of the tricuspid valve into the right ventricle. Consequently the tricuspid valve bisects the right ventricle and part of the right ventricle is incorporated into the right auricle, the valve itself is usually competent.

When the foramen ovale is completely closed, the blood from the right auricle flows into the right ventricle and is pumped out through the pulmonary artery to the lungs where it is oxygenated The oxygenated blood is returned by the pulmonary veins to the left auricle Thence it flows to the left ventricle and is pumped out by way of the aorta to the systemic circulation It is returned in the normal fashion by the superior and inferior venae cavae to the right auricle. There the cycle starts again

Clinical diagnosis The clinical findings vary with the severity of the abnormality of the tricuspid valve If the displacement of the tricuspid valve is slight symptoms are minimal Weakness and fatigue are the outstanding complaints If there is great displacement of the tricuspid valve the malformation leads to great cardiac enlargement The heart sounds are weak Murmurs are variable there may be a systolic murmur only or there may be a to-and fro murmur over the precordium Cardiac arrhythmias are common The electrocardiogram usually shows low voltage complexes of long duration in V_1 and V and normal deflection over the left precordium The liver becomes engorged but does not pulsate Tricuspid insufficiency is a late manifestation Terminally there may also be edema of the extremities Sudden death is not uncommon

DIAGRAM XIX–1

Head and upper extremities

R Lung

P.A.

S.V.C.

PV

Aorta

Pul. A

L Lung

Pul V

L A

Pulmonary Art.

R.A.

L V

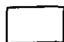
R.V.

Tricuspid Valve

I.V.C.

Descending Aorta

Trunk and lower extremities



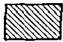
Arterial blood (fully saturated)



Venous and arterial blood
Cyanosis visible



Small admixture of venous blood
No visible cyanosis



Venous blood

CLINICAL FINDINGS

The clinical findings vary with the severity of the abnormality and the extent of the displacement of the tricuspid valve If the valvular abnormality is slight, the symptoms are minimal or absent and the condition is compatible with an active life The greater the displacement of the tricuspid valve into the right ventricle, the more inefficient is the action of the right side of the heart and the earlier and the more severe are the symptoms

Fatigue is more prominent than dyspnea Even though the patient may show persistent cyanosis and tire easily, there is little dyspnea The child is exhausted and rests but he is not short of breath He does not squat when tired

Palpitation is a common complaint, owing to the frequency of cardiac arrhythmias Many patients suffer from numerous extrasystoles or repeated attacks of paroxysmal tachycardia For this reason, in a person with an otherwise normal heart the persistence of a cardiac arrhythmia over a period of years should suggest the possibility of Ebstein s anomaly

The presence or absence of cyanosis depends upon the structure of the foramen ovale If, as previously mentioned, the valve covering the foramen ovale remains patent the valve will be forced open by the increased pressure in the right auricle and a right-to-left shunt will be established When a sufficient amount of reduced hemoglobin is shunted into the systemic circulation, the patient will develop persistent cyanosis The wider the patency of the foramen ovale, the greater is the shunt and the earlier is the development of cyanosis Although cyanosis may date from infancy, the development of cyanosis between two and five years of age is the rule rather than the exception

Polycythemia develops after cyanosis makes its appearance The red blood cell count, the amount of available hemoglobin, and the hematocrit reading gradually increase as the oxygen unsaturation of the arterial blood increases

The liver is enlarged and congested and frequently may extend to the umbilicus The force of the heart action is however, too weak and the alternations of the auricular and ventricular contractions which occur in the auricle and auricularized portion of the right ventricle are too ineffective to cause pulsations at the margin of the liver

CARDIAC FINDINGS

The heart may or may not be enlarged In the initial stage the right auricle is increased in size at the expense of the right ventricle Consequently the over all size of the heart is normal

If the tricuspid valve is markedly displaced into the right ventricle, the right auricle becomes a huge, ineffective chamber Since the right ventricle is not sufficiently large to receive all the blood from the right auricle, there is great dilatation of the right auricle and the pressure in that chamber gradually rises When the pressure in the right auricle exceeds that in the left auricle, if the valve covering the foramen ovale is not completely sealed, it will be forced open and a right-to-left shunt will be established As soon as the volume of reduced hemoglobin so shunted to the systemic circulation becomes sufficiently great, the patient develops cyanosis The course of the circulation is shown in Diagram XIX-2

PHYSIOLOGY OF THE MALFORMATION

The altered structure of the heart mainly affects the manner in which the heart functions The abnormal position of the tricuspid valve renders it difficult for the right auricle to expel its blood into the functional portion of the right ventricle Although normally the auricles contract before the ventricles, in Ebstein's anomaly the right auricle does not contract in the normal manner because part of the right ventricle lies above the tricuspid valve Consequently the upper or genuine auricular portion of the right auricle contracts before its lower portion, which is composed of ventricular muscle Thus with each auricular contraction, the blood in the upper part of the auricle is directed through the tricuspid valve to the lower chamber, but with each ventricular contraction, when the tricuspid valve is closed the blood in the lower part of the auricle cannot be directed into the functioning part of the right ventricle and is merely pushed back into the upper portion of the auricle This mechanism causes the right auricle to be extremely ineffective in directing blood to the functioning portion of the right ventricle Thus, with each heart beat, the lower part of the right ventricle receives less than its normal quota of blood It follows that with each ventricular systole only a small quantity of blood is pumped out into the pulmonary artery The pressure in the right ventricle is, however, normal and the pressure in the pulmonary artery is also normal It is the abnormal position of the tricuspid valve which causes the right side of the heart to be extremely inefficient The condition leads to progressive dilatation and hypertrophy of the right auricle and the right ventricle The dilatation is far greater than the hypertrophy, the right side of the heart becomes enormously enlarged and the pressure in the right auricle gradually rises As previously stated, the valve covering the foramen ovale may be forced open and a right to left shunt thereby established

DIAGRAM XIX–2

Ebstein's anomaly of the tricuspid valve and patency of the foramen ovale

The essential feature of this malformation is a congenital downward *displacement* of the tricuspid valve into the right ventricle. The result is that the tricuspid valve *bisects* the right ventricle and part of the right ventricle is incorporated into the right auricle. The tricuspid valve is usually competent.

The abnormal position of the tricuspid valve causes the right auricle to be greatly *enlarged* and the smaller lower portion of the right ventricle is the only part of that chamber which is of functional importance The greater the displacement of the tricuspid valve, *the larger* is the right auricle and the less effective is that chamber in the direction of blood to the functional part of the right ventricle.

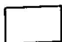
As the pressure in the right auricle rises, if the valve which covers the foramen ovale is not completely sealed, it will be forced open by the high pressure in the right auricle and a right to-left shunt will be established

Some blood from the right auricle flows into the left auricle and the remainder of the blood in the right auricle flows into the lower part of the right ventricle and is pumped slowly to the lungs where it is oxygenated The oxygenated blood is returned in the normal fashion to the left auricle, where it meets the blood which has been shunted from the right auricle into that chamber This mixture of oxygenated and venous blood flows into the left ventricle and is pumped out through the aorta to the systemic circulation and returned by the superior vena cava and the inferior vena cava to the right auricle. There the cycle starts again

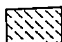
Clinical diagnosis The greater the displacement of the tricuspid valve the larger is the heart and the more probable it is that cyanosis is present. The patency of the foramen ovale acts as an escape valve for the high pressure in the right auricle but it also diverts the blood from the lungs and reduces the pulmonary blood flow Hence the lungs are exceptionally clear Weakness and fatigue are the outstanding complaints The heart is enlarged and often there is a to and fro murmur over the precordium and a gallop rhythm The liver is enlarged but does not pulsate The electrocardiogram shows low voltage curves of long duration in V_1 and V_2 and normal voltage in V_5 and V_6 Terminally there may be edema of the extremities Sudden death is not uncommon

Diagram xix–2

Head and upper extremities
R. Lung
L. Lung
S.V.C.
P.A.
P.A.
Aorta
PV
PV
Pulmonary Artery
L.A.
L.V.
R.A.
Foramen Ovale
R.V. upper chamber
R.V. lower chamber
I.V.C.
Descending Aorta
Trunk and lower extremities


Arterial blood (fully saturated)


Venous and arterial blood
Cyanosis visible


Small admixture of venous blood
No visible cyanosis


Venous blood

sternum, consequently there may be nothing characteristic in the size or shape of the heart, as illustrated in Figure XIX-4

Eventually (as shown in Figures XIX-5 and 6) the heart may be enormously enlarged both to the right and to the left The right auricle and the right ventricle may be so huge that they entirely obscure the left auricle and the left ventricle Even though the pulmonary arteries are of normal size, they are hidden behind the tremendous heart Because of the difficulty in the direction of the blood to the lungs, the lungs appear abnormally clear The hilar shadows are minimal and do not show expansile pulsations Whenever the *foramen ovale is not completely sealed* and the blood is shunted from the right auricle to the left auricle, the pulmonary blood flow is reduced Under such circumstances the lungs may appear so clear that the diagnosis of pulmonary stenosis may be erroneously entertained

Viewed in the left anterior-oblique position the heart may be flattened against the anterior chest wall for several inches, as shown in Figure XIX-5, and even when the patient is rotated at an angle of 60 degrees the left ventricle may

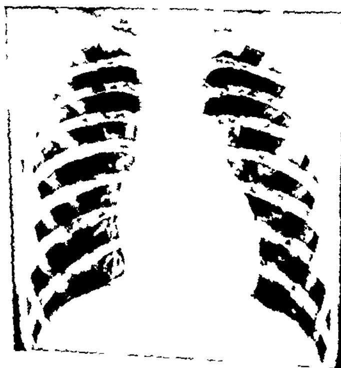


FIGURE XIX-4 Ebstein's anomaly with relatively slight displacement of the tricuspid valve (same patient as in Figure XIX-1) Young adult

The malformation leads to *progressive cardiac enlargement* The rate of cardiac enlargement varies with the severity of the abnormality of the tricuspid valve and the relative size of the two portions of the right ventricle The incorporation of the upper portion of the right ventricle into the right auricular chamber increases the size of this chamber but decreases the size of the functional portion of the right ventricle Furthermore, although the upper part of the right ventricle becomes incorporated into the right auricle, it does not contract simultaneously with the right auricle Thus first one part of the chamber contracts and then the other Only the upper portion of the right auricle contracts with auricular systole, the lower portion, which is composed of the "auricularized" part of the right ventricle, contracts with ventricular systole Furthermore, it is only with auricular systole that the blood flows through the tricuspid valve into the functioning portion of the right ventricle The tricuspid valve closes with ventricular systole Consequently the contraction of the lower part of the right auricle, which occurs with ventricular systole, cannot direct the blood to the lower chamber Thus the action of the right auricle is extremely inefficient, the anomaly leads to enormous dilatation of the right auricle and the auricularized portion of the right ventricle The cardiac enlargement extends both to the right and to the left of the sternum and eventually the heart occupies most of the thoracic cavity

The heart sounds are weak and of poor quality There is often a gallop rhythm *Murmurs* are variable A *systolic murmur* is the rule There may be a *diastolic murmur*, and not infrequently there is a *to and fro murmur* which may simulate a pericardial friction rub Indeed, it is the persistence of such a murmur over a long period of time, and its occurrence in the absence of other signs or symptoms of infection, which help to differentiate this murmur from a true pericardial friction rub

Cardiac failure develops insidiously and may become severe It is mainly right sided heart failure with engorgement of the liver and edema of the extremities Although tricuspid insufficiency has been considered so common in Ebstein's anomaly as to be almost synonymous with it, actually, except as a terminal event, its occurrence is rare

X RAY AND FLUOROSCOPIC FINDINGS

In the early stages, or when the displacement of the tricuspid valve is slight, x ray and fluoroscopic findings may be normal The portion of the right ventricle which becomes incorporated into the right auricle is hidden behind the

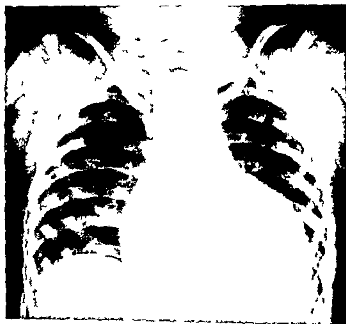


FIGURE XIX-6 Ebstein's anomaly with extreme displacement of the tricuspid valve (same patient as in *Figure XIX-3*) Young child

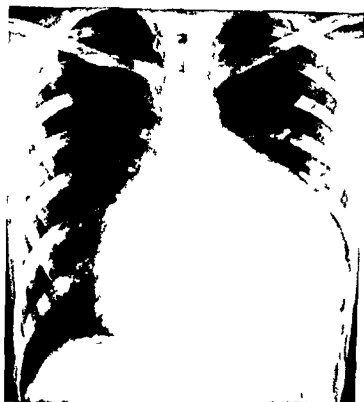
overlap the spine Under such circumstances the enlargement seen posteriorly is not due to a huge left ventricle but is caused by the backward displacement of the left ventricle by the huge right auricle and the right ventricle

Viewed in the right anterior-oblique position the heart appears to fill the entire chest cavity There is, however, no evidence of left auricular enlargement In both the anterior posterior position and the right anterior-oblique position, the esophagram is normal

The great enlargement of the heart and the weak pulsations cause the fluoroscopic appearance to simulate a pericardial effusion

ELECTROCARDIOGRAPHIC FINDINGS

The electrocardiogram, especially in older children and adults, may be a great aid in diagnosis Van Lingen and Bauersfeld[8] have shown that there is prolongation of both the auricular and the ventricular contractions The P waves are usually notched and of such long duration as to increase the P R interval Evidence of an intraventricular conduction disturbance is frequently seen in the standard leads The unipolar precordial leads give the clue to diagnosis The deflections in V_1 are of extremely low amplitude and of long duration The



Anterior posterior position



Left anterior-oblique position

FIGURE XIX-5 Ebstein's anomaly with marked displacement of the tricuspid valve (same patient as in Figure XIX-2) Child

whereas that in the right ventricle is normal The enormous size of the right auricle renders it difficult to pass the catheter into the right ventricle Moreover, catheterization may be extremely dangerous The auricularized portion of the right ventricle is so thin that there is danger that the catheter may pierce the wall In addition, the catheter may become entangled in the basket network of the tricuspid valve A further danger exists because of the susceptibility of patients with Ebstein s anomaly to cardiac arrhythmias In normal persons the manipulation of the catheter in the region of the tricuspid valve frequently produces arrhythmias Therefore it is not surprising that in patients with this malformation cardiac catheterization may initiate ventricular fibrillation or some other serious arrhythmia In the presence of tremendous cardiac dilatation prolonged auricular paroxysmal tachycardia may be fatal In addition, there is always the danger of cardiac arrest Therefore cardiac catheterization should not be undertaken lightly Nevertheless, Hernandez et al[9] have shown that intracardiac electrocardiograms taken during cardiac catheterization may be of great aid in diagnosis Normally the character of the electrocardiogram changes when the ventricle is entered In Ebstein s anomaly a ventricular type electrocardiogram may be obtained while the pressure curves indicate that the catheter is still in the auricle

Angiocardiography reveals prompt filling of the enormous right auricle and of the auricularized portion of the right ventricle, subsequently, as the lower portion of the right ventricle is filled, an even larger area becomes opacified (see *Figure* XIX–8) It is usually possible to see that the dye extends nearly to the margin of the cardiac silhouette, thus demonstrating the thinness of the right ventricular wall The dye lingers for a long time in the right auricle and only a small amount of blood and of contrast material is ejected into the lungs with each ventricular systole Consequently the lung fields are never well visualized The aorta may or may not be delineated It is never filled until after the left auricle has been visualized When there is a right to-left shunt through the foramen ovale, there may be faint early visualization of the left auricle, the left ventricle, and the aorta The concentration of the dye in the aorta seldom becomes great and the dye disappears from the aorta while it still lingers in the right side of the heart

DIAGNOSIS

Accurate diagnosis may be difficult. Cyanosis may or may not be present, it frequently becomes manifest when the patient is between two and five years of age Fatigue, not dyspnea, limits the child s activity The great right sided car

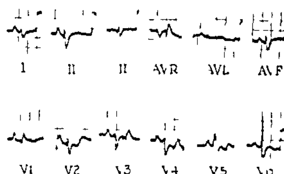


Figure xix–7 Ebstein's anomaly of the tricuspid valve (standardized at 1 cm 1 millivolt) (same patient as in Figure xix–2) Child

same may be true in V2 and occasionally even in V3 The deflections over the left precordium are, however, normal Figure xix–7 shows the characteristic electrocardiographic changes When present, such findings offer strong confirmatory evidence It is, however, worthy of note that the author knows of two young infants with Ebstein's anomaly whose electrocardiograms were entirely normal

In one instance the displacement of the tricuspid valve was so extreme that the infant lived but three days, nevertheless, the unipolar precordial leads all showed entirely normal voltage

Yater and Shapiro[2] have reported a case in which from time to time there were changes in the shape of the ventricular complexes They believed that these changes were due to the variability in the course of the excitation wave associated with the anomalous structure of the right ventricle

SPECIAL TESTS

The circulation time is usually prolonged Because of the difficulty in the expulsion of blood from the right auricle and the right ventricle, the circulation time may be exceptionally long Even if the foramen ovale is patent, only rarely is a sufficient amount of test material shunted into the left auricle and thence into the systemic circulation to give a short circulation time

The oxygen saturation of the arterial blood varies with the volume of the right to-left shunt If the foramen ovale is sealed, the oxygen saturation of the arterial blood is normal If the foramen ovale is widely patent, a considerable amount of reduced hemoglobin will be shunted into the systemic circulation and there will be a proportional reduction in the oxygen saturation of the arterial blood

Cardiac catheterization is of aid when both the right auricle and the right ventricle are catheterized The pressure in the right auricle is usually increased,

enlargement, the poor quality of the heart sounds, the blurred and confused murs, the presence of a to-and fro murmur, and the occurrence of severe iac failure with engorgement of the liver but without pulsations at its mar- suggest the diagnosis If the electrocardiogram shows low voltage in V_1 and nal deflections in V_5 and V_6, the diagnosis can be made with relative cer y The diagnosis is substantiated by the finding of a long circulation time by angiocardiographic evidence of a huge right auricle in which the dye ers for a long period Cardiac catheterization, if successful, shows an ele d pressure in the right auricle and a normal pressure in the right ventricle the intracardiac electrocardiogram shows a ventricular type curve in the er part of the auricle

ERENTIAL DIAGNOSIS

In the absence of cyanosis Ebstein s anomaly must be differentiated from te rheumatic fever, pericarditis, Lutembacher s syndrome, Fiedler s myocar s, and at times from persistent patency of the ductus arteriosus In the pres- e of cyanosis it closely resembles isolated valvular pulmonary stenosis and y require differentiation from a tetralogy of Fallot.

Acute rheumatic fever may be confused with Ebstein's anomaly when there systolic murmur at the apex transmitted to the axilla and also a to-and fro urmur over the heart. The absence of symptoms of acute rheumatic fever such fever, joint pains, rash, or abdominal pain and the finding of a normal sedi entation rate, as well as the clinical course, help to differentiate the two con ions

Pericarditis and pericardial effusion may be considered because of the to-and murmur and the enormous size of the heart. The long history of easy fatiga ity, the absence of other signs or symptoms of rheumatic fever or tuberculosis, d the knowledge that the condition dates from infancy or early childhood ch offers a clue to the correct diagnosis

Lutembacher s syndrome that is, an auricular septal defect combined with mitral stenosis may also be considered Patency of the foramen ovale in Ebstein's anomaly causes a reduction in the pulmonary blood flow The tremendous dila tation of the pulmonary artery and the conspicuous hilar dance so characteristic of Lutembacher s syndrome never occur in Ebstein's anomaly

Fiedler s myocarditis may be difficult to differentiate from Ebstein's anomaly In Fiedler s myocarditis murmurs are absent and a pericardial friction rub is rare In the chronic form of interstitial myocarditis, exacerbations and remissions are common

F

G

N